

Guidelines for Conducting Birth Defects Surveillance

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Introduction

In January of 1999, the National Birth Defects Prevention Network (NBDPN) established a Surveillance Guidelines and Standards Committee (SGSC) in order to develop and promote the use of standards and guidelines for birth defects surveillance programs in the United States. This set of guidelines is designed to serve as an important first step in the documentation of this process and as the vehicle for dissemination of the committee's findings.

The Guidelines for Conducting Birth Defects Surveillance (henceforth referred to as The Surveillance Guidelines) were developed with three major long-term objectives in mind:

- > To improve the quality of state birth defects surveillance data, including accuracy, comparability, completeness, and timeliness.
- > To enhance the utility of state birth defects surveillance data for research on the distribution and etiology of birth defects.
- To encourage and promote the use of state birth defects surveillance data for the purposes of linking affected children with services and evaluation of those services.

The technical guidelines that make up this document provide a way of improving the quality of birth defects surveillance data, which in turn enhances their use in support of the latter two objectives. Fundamental to quality is ensuring that procedures for all aspects of data definition, collection, management, and analysis are established and followed. Because state-based surveillance systems operate with different objectives and data needs, it is clear that, with respect to procedures and standards, "one size does not fit all." It is also clear, however, that common guidelines can provide a basis for the development of system-specific operating procedures and supporting manuals.

Variation among surveillance programs is manifest along several dimensions. These include:

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- *Objectives*, which can be very diverse but commonly include:
 - Providing baseline data on occurrence
 - Identifying populations at increased risk
 - Monitoring changes in occurrence
 - Investigating clusters
 - Collaborating with research
 - Estimating service needs
 - Referring affected children to services
 - Evaluating prevention programs
- > Case ascertainment methods
 - Active case finding
 - Passive case reporting
 - Combined

- > Organizational location
 - Health department
 - University
 - Other

The first two dimensions – objectives and case ascertainment methods – are of particular significance in attempting to develop guidelines that have the breadth to be useful (i.e., universality), while at the same time making clear that there is not necessarily a common denominator across programs. Thus most of the guidelines in this volume are phrased as *recommendations* or "shoulds," as opposed to *standards*, which could be interpreted as "musts." The exception to the latter is Chapter 10, which refers the reader to information on how data are to be reported to NBDPN for the Annual Report. The relevance of organizational location to the guidelines is probably restricted to legislative issues, which are addressed in Chapter 2.

The Surveillance Guidelines consist of a series of chapters covering the fundamental aspects of developing, planning, implementing, and conducting surveillance for birth defects and using the resulting data. Although the focus is on birth defects, most of the principles described are relevant and applicable to surveillance for any health outcome. Just as the methods and strategies developed for birth defects in the Metropolitan Atlanta Congenital Defects Program provided a blueprint for the subsequent development of the Metropolitan Atlanta Developmental Disabilities Surveillance Program, the information included in these guidelines can provide a blueprint for the development of surveillance for developmental disabilities among the states.

On reviewing the guidelines, the reader will note that a number of the chapters are supported by appendices. In many instances these appendices are designed to provide additional information on technical issues considered. In some cases they provide extensive detail on procedures that are currently being used by surveillance programs. Because of their size, three documents cited as appendices will only be available in electronic format. These are the NBDPN Abstractor's Instructions (Chapter 3, Appendix 3.2) and the Texas Disease Index and the CDC Six-digit Codes (Chapter 5, Appendices 5.1 and 5.2, respectively). Information on how to access the electronic format is included in each appendix.

The Surveillance Guidelines are being published in two formats: as print copy and through the NBDPN website. The Surveillance Guidelines and Standards Committee anticipates updating and revising the guidelines over time. Whenever a revision is published, a revision date will appear in the chapter header to distinguish that page or pages from previous versions. Because we anticipate this will be a living document, we encourage comments, suggestions, and corrections. If you have such, please submit them through the link to the Surveillance Guidelines and Standards Committee on the NBDPN website.

This set of guidelines represents a great deal of work by a large number of individuals. The development of the document was carried out by the NBDPN Surveillance Guidelines and Standards Committee. A working group for each of the chapters did most of the writing. When chapters were completed in draft form, they were submitted to the SGSC Steering Group for review and suggested revisions. When a draft was considered acceptable to the Steering Group it was sent to Dr. Lowell Sever of Battelle Centers for Public Health Research and Evaluation, the editorial consultant for the reference manual. Dr. Sever then edited the chapter, returning it to the Steering Group, and working groups when necessary for clarification and acceptance of his revisions. Several of the chapters were also sent to specially assembled "Focus Teams" for review and assessment of the technical content. When the final content was agreed upon, the chapter was submitted to a Battelle technical writer and editor for finalization of structure and format.

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We have compiled all of the contributors to this intensive process into a single acknowledgements page. *The Surveillance Guidelines* represent a significant and complex undertaking that could not have been accomplished without the contributions of this large number of individuals, and we thank them all.

We dedicate this milestone document to Larry Edmonds of the Centers for Disease Control and Prevention in recognition of his strategic vision, inspiring leadership, and steadfast commitment – both to the National Birth Defects Prevention Network and to the enhancement of birth defects surveillance generally – throughout the remarkable process of developing *The Surveillance Guidelines*.

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Chapter 1 The Whys and Hows of Birth Defects Surveillance – Using Data

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1.1 Introduction

The ultimate value of any public health surveillance program lies in the ways in which the data collected are used to improve the health of the public. State birth defects surveillance programs are no exception; they exist to improve public health. Every program must have clear goals and objectives that drive how their surveillance data are used toward improving public health. Public input through partnerships with state agencies and organizations and the effective utilization of advisory committees are essential to establishing and revising program objectives and ensuring that the resources exist to meet them.

The purposes and objectives established by state birth defects surveillance programs are constantly evolving. Some objectives are traditional, such as those having to do with the epidemiologic purposes of surveillance; others have emerged more recently, serving to broaden the scope of surveillance programs. Birth defects surveillance programs increasingly use data for services planning and evaluation, for development and evaluation of prevention strategies, to inform parents of children with birth defects about available services, for studies of the societal impact of birth defects, for referral of families to needed services and resources, and for clinical research studies. The consistent theme among these emerging data uses is how birth defects surveillance may benefit other programs in the quest to improve the public's health. In the face of fluctuating resources for public health and obstacles resulting from concerns about confidentiality of health records, the need to incorporate public input in planning and priority-setting has never been greater. This chapter will attempt to address some of the issues in the forefront as we plan for the future of birth defects surveillance.

In the remainder of this chapter we present the rationale for conducting birth defects surveillance (Section 1.2), key steps in establishing a state-based birth defects surveillance program (Section 1.3), and some important uses for birth defects surveillance data (Section 1.4). References cited in this chapter may be found in Section 1.5.

Chapter 1 1-1 Using Data

1.2 Rationale

When contemplating initiating or enhancing a birth defects surveillance program, a number of questions come to mind:

- What is the rationale for conducting birth defects surveillance?
- ➤ Why is birth defects surveillance important?
- ➤ How do birth defects surveillance data benefit other programs?
- ➤ What are the barriers to collection and full utilization of birth defects surveillance data?

In this chapter, we provide answers to these questions, which may help you advocate for and prepare to launch or expand a birth defects surveillance program in your area.

1.2.1 What is the rationale for conducting birth defects surveillance?

CDC defines public health surveillance as:

The ongoing, systematic collection, analysis, and interpretation of health data essential to the planning, implementation and evaluation of public health practice, closely integrated with the timely dissemination of these data to those who need to know. The final link of the surveillance chain is the application of these data to prevention and control. A surveillance system includes a functional capacity for data collection, analysis, and dissemination linked to public health programs (Centers for Disease Control, 1988).

It is clear from this definition that a birth defects surveillance program must establish goals and objectives for how data are to be collected, analyzed, disseminated, and used. It is through the latter (i.e., data use) that the efforts from the former are translated into public health action and health improvement. Thus, using data to meet a program's objectives is the most important aspect of any public health surveillance program; merely collecting data is not enough. How data are being used is also what programs tout when they need to showcase their activities to agency officials and legislators.

Because of the essential relationship of the ultimate uses of data to the design and conduct of birth defects surveillance, we begin these guidelines with a consideration of fundamental data-related issues, considering not only the rationale for birth defects surveillance but the key steps for establishing state-based birth defects surveillance programs, followed by a discussion of the use of surveillance data for improvement of the public's health. Every surveillance program should have a plan for data utilization that incorporates public input on all phases of the process – from data development, through data collection, to data dissemination to the public. Suggestions for developing a data utilization plan are presented in Section 1.2 below.

1.2.2 Why is birth defects surveillance important?

States have many reasons for conducting birth defects surveillance. The value of birth defects surveillance lies in how the data are collected and how they are used, with respect to the goals of the program. All programs should establish goals and objectives, which make it clear that the ultimate rationale for conducting public health surveillance is to have data that can be used to improve the health of the public. Reporting the

data certainly qualifies as "using the data," yet this should never be considered sufficient as it fails to meet the definition of public health surveillance cited above.

The objectives of state birth defects surveillance programs have evolved over the past 40 years. Lynberg and Edmonds (1992) assessed the objectives that had been established by surveillance programs by the early 1990s. Table 1.1 organizes these objectives under broad purposes originally suggested by Reed and Meaney (1988) with some slight modifications. A review of the table highlights the potentially broad mission of birth defects surveillance, providing state programs with a way of assessing how they are utilizing data currently and possible new uses.

Table 1.1 Purposes and Objectives of Birth Defects Surveillance

Purposes	Objectives
Epidemiologic	Develop timely baseline birth defects rates
	Monitor trends and relationships to environmental factors
	Perform cluster investigations
	Provide basis for ecologic and etiologic studies
Planning/Prevention	Provide data for services planning
	Provide basis for prevention strategies
	Evaluate efficacy of preventive services
Educational/Social	Inform public about public health importance
	Inform parents about resources and care facilities
	Provide data for studies of economic impact
	Provide data for follow-up studies of long-term effects
Healthcare and human services	Refer children to services and resources
	Evaluate services utilization
Clinical	Provide basis for clinical research

Adapted from Lynberg and Edmonds (1992) and Reed and Meaney (1988) with modifications.

1.2.3 How do birth defects surveillance data benefit other programs?

The benefits of birth defects surveillance data to human service programs include: identifying children in need of services to ensure that they and their families are referred appropriately; evaluating service utilization by children with birth defects and their families; and planning the location of services for particular conditions in areas of highest frequency. An important use of surveillance data is monitoring birth defects trends following the initiation of prevention programs in order to evaluate their effectiveness.

One of the public health benefits of the computer age is enhanced capacity for record linkage. Record linkage using public health data has a longer history than most people realize, beginning in the 1950s with the availability of computers in university settings. Pioneering investigators like Harold B. Newcombe (1962) recognized the utility of linking vital records data in studying human populations. The potential now exists for extensive computerized record linkage in birth defects surveillance programs, allowing for the tracking of children with a health-related condition from the point of identification through access to services. Many computer-based systems already exist for documenting health care delivery, including diagnostic and procedure codes. Birth defects surveillance records have been linked to many other public health program databases. These include, for example, newborn screening to conduct epidemiologic studies, special education data to predict the need for services for children with mental retardation, and early intervention

program data to assess the overlap and utility of a birth defects surveillance program as a "child find" resource.

In the final section of this chapter we describe a number of applications of these approaches that can serve as models for states developing birth defects surveillance programs, as well as for programs considering expansion of the current uses of their data. To date, the potential for applications of these types exceeds available resources to support them and to overcome some of the obstacles discussed immediately below.

1.2.4 What are the barriers to collection and full utilization of birth defects surveillance data?

While improved methods and technological advances have increased our ability to collect data, there have been intensified efforts to protect the confidentiality of records and the information they contain. Many birth defects surveillance programs – based both in health departments and in other institutions such as universities – have encountered increasing concerns and pressures as a result of Health Insurance Portability and Accountability Act (HIPAA) regulations and issues surrounding their interpretation and implementation. A variety of HIPAA-related issues are discussed in Chapter 2 of these guidelines. Even though the HIPAA regulations include public health exclusions regarding access to records without a patient's consent, programs have seen increased awareness and concerns on the part of hospitals and clinics reporting cases and data. These concerns are magnified when a surveillance program attempts to expand data usage through linkage to other databases covered under HIPAA regulations.

Prior to HIPAA, concerns often surfaced about data sharing among officials in different programs within the same state agency or among programs located in different agencies. Such concerns were usually due to program-specific regulations about data use. Program regulations frequently impede attempts to link records between case-finding databases and service-delivery databases. As a result, attempts to meet the very reasonable public health goal of ensuring access to services by those in need may be thwarted. Thus, programs are strongly urged to consider strategies for surmounting these problems well in advance of undertaking data collection and record linkage.

Chapter 1 1-4 Using Data

1.3 Synopsis of Key Steps in Establishing State-Based Birth Defects Surveillance Programs

In this section we outline some of the key steps in establishing a birth defects surveillance program. These include:

- ➤ Defining the objectives and purposes of the program
- Considering legal issues
- > Engaging external support
- ➤ Leveraging resources
- Considering record linkage

Time devoted upfront to serious consideration of these issues will be well spent and will ensure that the resultant program is established on a firm footing.

1.3.1 Defining the Objectives and Purposes of the Program

The success of a birth defects surveillance program is likely to be highly dependent on the host agency's commitment and support. Without programmatic commitment and resource support at the agency level, programs are apt to languish in circumstances that do not allow much beyond the collection and reporting of data. In these situations, using data in ways other than the calculation of rates and their dissemination in reports is usually not possible. Programs committed to expanding how birth defects surveillance data are used must establish programmatic objectives and demonstrate to agency officials how the data could be used. This involves prioritizing what uses would be of greatest utility in terms of meeting agency goals and objectives, demonstrating (or "marketing") to the agency how beneficial these data uses could be, and working to achieve commitment of additional agency resources.

Another strategy for increasing support from the agency in which the surveillance program resides is to gather support from other intra-agency programs and from external agencies that could benefit from the use of birth defects surveillance data to meet their own programmatic goals. Often other programs and agencies, given enough information about birth defects surveillance and the objectives of the program, will see potential uses of the data that are beyond the current scope of the surveillance program.

There has been an increase in intra-agency collaboration during the last ten years through the availability of federal support for data linkage and integration. A prime example of data collaboration would be linking birth defects surveillance databases with Children with Special Health Care Needs (CSHCN) program databases that collect data on program enrollment and services. These linked data sets could then be used to evaluate the rates at which this long-term maternal and child health program is utilized. Such applications have been accomplished in some states through grant support from the Maternal and Child Health Bureau (MCHB) of the Health Resources and Services Administration (HRSA) and through cooperative agreements with the Centers for Disease Control and Prevention (CDC). Interagency collaboration in linking birth defects surveillance program databases with services databases (such as those for early intervention programs or developmental disabilities) have begun in a few states. The benefits to be gained in this way – i.e., by utilizing birth defects surveillance data as a means of identifying children eligible for special programs, such

as early intervention – is clearly a "selling point" that can lead to additional resource allocation, either from within the host agency for the birth defects program or from an external agency in need of the data.

Most birth defects surveillance programs experience cyclical problems with availability of state resources, leading them to define precisely what they can and cannot do given the resources available to them. While it is certainly necessary for programs to realistically budget their resources to ensure continued viability, programs also need to engage both intra-agency and interagency support for their goals and objectives as a means to maintain and expand a surveillance program. At a minimum, programs should allocate personnel time to educate officials of their own agency and other agencies about birth defects surveillance and its importance and potential uses in the public health field.

1.3.2 Considering Legal Issues

To the extent possible, programs should consider the inclusion of references to data use in the legislation that authorizes birth defects surveillance. Given the relative ease with which rules – as compared to laws – can be changed, it is generally desirable to make references to potential data uses for surveillance data more general in the statute and more specific in the rules. Rules and regulations that refer to the authorizing statutes are the obvious choice as to where best to specify detailed uses to which surveillance data will be put. Relevant issues and legal considerations are discussed extensively in Chapter 2 of these guidelines.

1.3.3 Engaging External Support

Beyond seeking intra-agency and interagency support for a new surveillance program or for expansion of an existing surveillance program, program staff should also seriously consider means to attract the support of both non-governmental partnering organizations and the public.

Partnering organizations. The importance of building partnerships with organizations such as the local March of Dimes can never be sufficiently stressed. In recent years, the success story of the birth defects surveillance program in North Carolina is arguably without peer. The program has consistently credited the partnership it built with the March of Dimes as a major contributor to its success in garnering additional resources for the program. In Texas, the March of Dimes was also instrumental recently in restoring funds to maintain the Texas Birth Defects Monitoring Division, funds that had not been requested in the budget put forward by the Texas Department of Health. These kinds of partnerships should be entered into with clear and consistent agreement among the players regarding the objectives of the program relative to data usage, prioritization of data uses, and planning toward future applications of the data. In other words, the contribution of organizations such as the March of Dimes can be beneficial from the design of data utilization plans through to the reporting of actual outcomes.

Advisory committees with agency, organizational, and public representation, including political officials, are another means of obtaining input regarding uses of birth defects surveillance data. The available computer technologies such as listservs and webpages decrease the need for face-to-face meetings among interested parties, while increasing the frequency with which information about a program can be communicated and feedback solicited. New ideas about potential uses to which a program's data can be put and the resources needed to accomplish programmatic activities can be shared with advisory committee members for immediate feedback as to the feasibility of the idea and its potential for success.

Programs should create opportunities for formal input from advisors on a regular basis to ensure the availability of support in times of fiscal crises. Advisory group members' knowledge of surveillance data collection activities and uses for surveillance data can be critical to securing resources for a program in times when limited resources require justification for program continuation.

Public involvement. Birth defects surveillance programs generally have not engaged consumer and parent participation other than through advisory group representation. Members of the public, including parents of children identified through these programs, are often not well informed about public health surveillance activities. If not already doing so, birth defects surveillance programs should engage both consumers – here defined as adults with birth defects, and parents and caretakers of children with birth defects – in the planning and implementation of any and all programmatic changes. There are a number of advocacy and parent support groups, such as the Spina Bifida Association of America, Family Voices, and the Alliance of Genetic Support Groups, that can play important roles in planning and conducting birth defects surveillance programs.

Programs should embrace the concept of *participatory action research* (PAR) (Whyte, 1991). PAR is a way to obtain public input into programmatic activities from design though dissemination of results. PAR ensures input from the community members most affected by potential data uses. Again, as discussed with respect to advisory group input, computer technology can be immensely beneficial in obtaining feedback on new initiatives and more importantly in soliciting input about programmatic activities from community members.

1.3.4 Leveraging Resources

For birth defects surveillance, as for other public health surveillance programs, the ways in which data are used will influence continued availability of program resources. In the age of evidence-based medicine and increased emphasis on demonstrating program efficacy for continued support, birth defects surveillance programs should work toward expanding data use. Fiscal trends in states suggest that the likely survivors in times of increasingly fewer tax-based resources will be programs that adapt by reinventing themselves in terms of data utilization. While emphasizing the application of surveillance data to improving human services and then evaluating their impact will not ensure the survival of a program, it should increase its chances.

Surveillance programs (particularly those housed in health departments) may be given adequate resources for data collection and management, but often do not have adequate personnel or resources for data analysis beyond simple descriptive reporting. Program managers and staff often use lack of adequate resources as an excuse to minimize the number of new initiatives they undertake, but this may well be a short-sighted approach. We have already discussed the importance of partnerships, advisory groups, and public involvement in increasing the probability of acquiring additional resources. While programs must, realistically, work within the limits of available resources, partnerships with agencies and institutions can represent a means to extend and enhance programmatic achievements. Universities, particularly those with public health training programs or medical schools, will have faculty and trainees potentially interested in birth defects. What a birth defects surveillance program lacks in resources for data analysis and research often can be compensated for through partnerships with interested faculty members willing to direct student theses and dissertations that focus on birth defects. New programs and programs that do not currently have such partnerships should give serious consideration to forming these types of collaborations, which can lead to additional resources through contracts and grants.

1.3.5 Considering Record Linkage

As touched upon in Section 1.3.1, the potential to link records and consolidate information from different databases contributes to the public health applications of surveillance data. For example, data from birth defects surveillance programs can be used to determine whether reported cases of birth defects represent existing cases in other databases, such as records in interdisciplinary clinics and schools with programs to assist children with disabilities. The ability to link records on individuals in more than one database can

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streamline the treatment and referral processes and help maintain a certain level of fidelity and trust in prevalence data. Record linkage can streamline the research process by consolidating several different databases. Another utility of record linkage is the ability to supply crucial data required for various research efforts. Specifically, the data located in one database can be used to elicit information from a second.

Chapter 1 1-8 Using Data

1.4 Uses of Surveillance-based Birth Defects Data

Most US states have implemented birth defects surveillance programs that monitor and disseminate information regarding birth defects. Public health staff and researchers nationwide have used these data in a variety of ways. The actual and potential uses of birth defects data are discussed and exemplified in the following sections. Data from birth defects surveillance programs can be employed to define the magnitude of a problem, to support research, as well as to assess the efficacy of prevention and treatment, playing a key role in the core public health function of *assessment* (Institute of Medicine, 1988).

For convenience, the uses of birth defects surveillance data can be grouped into the following categories:

- Prevalence studies
- > Epidemiologic studies
- Mortality assessment
- > Needs assessment for services
- > Referral to clinics and services
- Program evaluation
- > Clinical research

Each of these categories of use will be discussed in further detail below. While comprehensive coverage of works in each of these categories is beyond the scope of this chapter, we have selected published studies that exemplify the kinds of research that can be conducted in each category. Naturally, what an individual program is able to do depends ultimately on its goals and objectives. When programs are faced with limited resources to conduct data analysis and research, collaborations with universities or contractors with epidemiologic expertise can often yield mutually satisfactory results.

1.4.1 Prevalence Studies

A common use of data produced by birth defects surveillance programs is to describe the occurrence (*prevalence at birth*) of the monitored conditions. Such uses of surveillance data include identification of trends in birth defects occurrence, definition and evaluation of clusters of congenital defects, and assessment of the need for resources and interdisciplinary services.

Khoury et al. (1986) is an example of an early study by a state surveillance program that used data in this way. This study was the outcome of a partnership between the state health department-based surveillance program and university-based researchers. Khoury and co-workers used 1984 data collected from the Maryland Birth Defects Reporting and Information System (BDRIS) to determine rates of occurrence and to identify potential trends. The prevalence at birth of "sentinel" defects, as determined from the Maryland BDRIS data, was 52.7 per 10,000 qualified births. Furthermore, trends in the occurrence of several specific birth defects were identified. The study revealed an association of low birth weight and prematurity with birth defects, an association between twinning and the rate of birth defects, racial differences in the prevalence of neural tube defects, and a relationship between Down syndrome and advanced maternal age. The importance of determining prevalence at birth is that the data can be compared with similar data collected from other birth defects monitoring systems to assess differences in rates that may exist among

surveillance areas and to direct further research efforts in an attempt to identify the reasons behind the differences

An example of a more recent prevalence study is one reported by Ethen and Canfield (2002), who investigated the effects of including elective pregnancy terminations, prior to 20-weeks gestational age, on birth defects prevalence. In many surveillance programs, pregnancies ending prior to 20 weeks gestational age, including elective terminations, are not ascertained to be included among reported cases. The researchers concluded that when elective terminations at less than 20 weeks were considered, the prevalence of some congenital defects increased, while others remain unchanged. Specifically, anencephaly, spina bifida, and encephalocele increased substantially, while cleft palate did not change. The underlying assumption is that pregnancies resulting in debilitating or potentially terminal conditions are more likely to be terminated electively than those resulting in less severe or treatable malformations.

These two studies show the potential usefulness of prevalence data to reveal important trends and associations. These types of data often provide the impetus to initiate subsequent research. A consequence of producing birth defects prevalence data is that it frequently opens other avenues of exploration. Quite simply, without basic prevalence data to lead inquiry, many research investigations never would be conceptualized, much less carried out.

1.4.2 Epidemiologic Studies

Cases from birth defects surveillance programs have played key roles in conducting etiologic research in the United States and internationally. Cases from the Metropolitan Atlanta Congenital Defects Program (MACDP) have provided the basis for numerous research studies that have shed light on both the causes (Khoury et al., 1982; Oakley, 1984; Erickson, 1991; Dott et al., 2003) and prevention (Roberts et al., 1995; Olney et al., 2002) of birth defects. Similarly, the California Birth Defects Monitoring Program (CBDMP) has been the source of cases and etiologic research that has resulted in dozens of seminal papers on a variety of specific congenital malformations and their risk factors (Croen et al., 1991; Shaw et al., 1996; Ritz et al., 2002). Other state programs have contributed cases for epidemiologic studies leading to a growing number of multi-state investigations of specific risk factors (for example, Olney et al., 1995). Reference to the annual report of the International Clearinghouse for Birth Defects Monitoring Systems (International Centre for Birth Defects, 2002) demonstrates the large number of studies based on individual surveillance systems and collaborative projects among programs.

An example of an early methodological study, based on surveillance data, is a study by Khoury et al. (1988) that assessed the patterns of maternal residential mobility between conception and delivery. The authors' rationale was that most epidemiologic studies of environmental risk factors are based on maternal residence at the time of delivery. Such an assessment would be invalid, however, in instances where the mother had moved prior to delivery. The researchers examined demographic data for infants born with congenital defects. Both the demographic data as well as the birth defect data were taken from the Maryland BDRIS in 1984. The researchers concluded that, on average, 21% of all mothers whose pregnancies resulted in a child affected by one of the birth defects included in the Maryland BDRIS had moved between conception and delivery. This is important for several reasons. First, it is well understood that the effects of environmental teratogens occur early in embryogenesis; so assessing the influence of environmental exposures must be related temporally to conception. In addition, potential exposures to teratogenic environmental factors could possibly be misrepresented if examined at delivery rather than around the time of conception. Maternal mobility could also skew data regarding geographic clusters of birth defects. This study was made possible because the Maryland BDRIS determines the residence of the mother not only at the time of delivery, but also at the time of conception. This is an important aspect of the Maryland BDRIS that is not common to all birth defects surveillance programs.

Examples of surveillance-based etiologic research of associations between maternal exposures and congenital defects include studies of cigarette smoking and orofacial clefts. Among the earliest research efforts investigating this association was a study by Khoury et al. (1987) using data collected in 1984 from the Maryland BDRIS. A case-control study examined the history of cigarette smoking among mothers of infants with orofacial clefts and a group of control mothers. The researchers concluded that odds ratios for cleft palate (2.39, CI 1.04-5.45) and cleft lip with and without cleft palate (2.56, CI 1.13-5.78) were increased for women who smoked. Furthermore, the researchers identified a dose-response effect. Khoury and his coworkers also took into account possible confounding factors, including race, gender, residence, maternal age, parity, and several pregnancy exposures or complications. None of these affected the results significantly. This is a classic example of how surveillance-based birth defects data can be used to examine etiologic factors through the use of simple epidemiologic techniques. Sometimes the importance of earlier epidemiologic studies is not appreciated when comparing them to more recent research. It is worth noting that the association between maternal cigarette smoking and orofacial clefts has been corroborated through more recent studies using several surveillance-based investigations. The paper by Khoury et al. (1987) has been cited in many contemporary research publications (Shaw et al., 1996; Lieff et al., 1999).

Some states have used surveillance data to look for associations between environmental factors that are known to cause specific birth defect syndromes and other birth defects. For example, maternal alcohol use during pregnancy is a known cause of the fetal alcohol syndrome, but its role in more common, isolated, craniofacial defects is not well understood. A population-based, case-control study of orofacial clefts was conducted in Iowa based on births from 1987-1991 (Munger et al., 1996). Cases were identified by the Iowa Birth Defects Registry and classified as having a cleft lip with or without cleft palate (CLP) or cleft palate only (CP) and as to whether the cleft was isolated or occurred with other birth defects. Controls were selected from normal Iowa births. Maternal alcohol use during pregnancy was classified according to self-reported drinks consumed per month. Compared to women who did not drink alcohol during pregnancy, the relative odds of isolated CLP rose with increasing level of maternal drinking as follows: 1-3 drinks per months, 1.5; 4-10 drinks per month, 3.1; more than 10 drinks per month, 4.7 (chi-square test for trend, P = 0.003). Adjustment for maternal smoking, vitamin use, education, and household income did not substantially alter the results. No association was found between alcohol use and isolated cleft palate or clefts in children with multiple birth defects. Based on these data, alcohol use during pregnancy may be a cause of isolated cleft lip with or without cleft palate.

As described, epidemiologic investigation is an important area of research supported by birth defects surveillance data. In the past, this research effort primarily focused on environmental exposures as possible etiologic factors. However, with the recent explosion of molecular genetics and a more thorough understanding of molecular biology, the avenues of epidemiologic investigation have widened significantly. Investigators now have an enhanced ability to examine the contributions of both maternal and fetal genotypes to disease risk. Examination of the interplay between genetic predispositions/susceptibilities and environmental exposures is a growing area of study, with potential major implications with respect to understanding birth defects etiology. This is illustrated by the genetic component of the National Birth Defects Prevention Study, a multicenter case-control study being conducted by CDC and participating state surveillance programs (Yoon et al., 2001; Rasmussen et al., 2002).

Continuing with the study of the association between smoking and clefts, epidemiologic studies have focused on the relationship between certain alleles of a transforming growth factor and maternal cigarette smoking with regard to risk of orofacial clefts. The most promising associations are seen in polymorphisms of the transforming growth factor alpha (TGF α) gene taq1 and maternal cigarette smoke exposure. An example is a study by Hwang et al. (1995), supported by surveillance data, that examined this association. The data on infants born with orofacial clefts were taken from the Maryland BDRIS. The Maryland BDRIS was not only able to supply cases of orofacial clefts, but also information about maternal prenatal behaviors, including maternal smoking during pregnancy. Cases were genotyped and screened for the rare C2 taq1 polymorphism.

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The researchers concluded that the C2 genotype, combined with maternal smoking, significantly increased the risk of orofacial clefts. Using data collected through a birth defects surveillance program, they were able to identify a possible interaction between an environmental exposure and a genetic predisposition with respect to risk for orfacial clefts.

Studies like this represent another generation of epidemiologic research. The power of these molecular epidemiologic studies lies in their ability to elicit possible etiologies of birth defects beginning with prevalence data, demographic information, and biologic samples. While the epidemiologic research methods have evolved significantly, the ultimate goal of these studies has remained constant: namely, to identify, define, and associate birth defects with possible etiologic factors. The development and application of molecular genetic methods serve as stepping stones to future research based on surveillance-derived cases.

1.4.3 Assessing Mortality Associated with Birth Defects

A 1995 Texas study assessed survival rates for selected birth defects among babies born between January 1, 1995 and December 31, 1997, by linking two databases: the state's active birth defects registry and the infant death registry (Nembhard et al., 2001). The goal of the study was to determine mortality among cases with various birth defects identified through the birth defects surveillance system by matching those cases against infant death files. Specifically, the researchers found the birth defects with the lowest survival were anencephaly (0%) and trisomy 13 (7.4%), while the birth defects with the highest survival were gastroschisis (92.9%) and trisomy 21 (92.3%). These survival data were only for the first year of life.

Another example of a mortality study is that carried out by Druschel et al. (1996), who examined infant mortality among children with orofacial clefts, comparing their mortality rates to those of children with no congenital malformations. In the absence of malformations in other organ systems (isolated clefts), mortality was not increased among children with orofacial clefts. The study revealed, however, that many children with orofacial clefts have other malformations that increase their risk of death. These findings suggest the need for careful evaluation of possible additional malformations among children with orofacial clefts as these children may be at higher risk of death.

1.4.4 Estimating the Need for Services

Estimating service needs based on birth defects prevalence has significant direct social consequences. Accurately predicting the demand for various interdisciplinary clinics and social and educational services is critical for children born with birth defects. Estimating future service needs allows for capacity building to ensure that necessary resources will be accessible and that appropriate professionals will be available to provide the services.

Brewster et al. (1992) linked demographic and diagnostic data from 1980 – 1982 in a birth defects surveillance program database (the Arkansas Reproductive Health Monitoring System) with education databases. The data were first used to estimate the percentages of infants with specific birth defects who were at risk for developmental disabilities and mental retardation. Once prevalence rates were determined, two clinicians estimated the various services that would be needed by children with the various birth defects most likely to contribute to developmental disabilities. This included academic and other services these infants would require as they matured. The researchers estimated that between 32% and 56% of all children in schools who were classified as mentally retarded were also identified by the Arkansas Reproductive Health Monitoring System.

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This study showed that recognition of children with mental retardation, who were also identified years earlier as having congenital defects, allowed researchers to refine their estimates of the birth defects that will contribute most significantly to mental retardation in school-aged children. This is useful in improving the ability of health care professionals to predict accurately future needs of the current cohort of newborns with birth defects

1.4.5 Referral to Services

Information collected as part of birth defects surveillance can be used to refer specific children and their families to appropriate services. Established referral networks serve as a resource for children and their families to learn about available medical services, community programs, and social support. Affected children and their families can be connected with appropriate services in a timely fashion.

Many papers have been written detailing the process of identification and ultimate service referral. One of the first papers on this topic comes from the Maryland BDRIS, where investigators examined the referral of children identified with orofacial clefts through the surveillance program to the Maryland Crippled Children's Service Program in the 1960s (White, 1981). This study examined referral rates to services. A more recent paper on referral and treatment patterns for orofacial clefts comes from Florida, where referral and treatment patterns of live-born Florida infants diagnosed with orofacial clefts identified through the Florida Birth Defects Registry were determined (Williams et al., 2003).

Another example is a paper describing service referrals in Colorado that use birth defects data taken from their birth defects surveillance program (Montgomery and Miller, 2001). The Community Notification and Referral Program (CNRP), operating from within the state's health department, uses birth defects data to link affected infants with an organization that can refer them and their families to agencies and interdisciplinary clinics. In 1998, 259 families were referred for services as a result of being identified through the birth defects registry. There are a number of services to which patients are commonly referred, including developmental screening and evaluation, public health programs, early intervention programs, financial assistance, parenting classes, medical services, recreational programs, and family support groups. Additionally, the effectiveness of this program has been assessed through the use of surveys and questionnaires.

A review of the use of surveillance data relative to provision of early intervention services can be found in a recent paper on identification and referral programs by Farel and colleagues (2003). Having agencies use birth defects data to link patients with appropriate services is a critical data use that has immediate and direct impact on the lives of those affected. Although epidemiologic and laboratory efforts may illuminate etiologies and possible preventive measures for future use, the fact remains that effective therapeutic efforts in the present can significantly improve the lives of persons with birth defects; scientific studies take years to complete and primarily aid future patients. Meanwhile, there are people who require immediate assistance, and service referral is an important mechanism through which they can receive that help.

1.4.6 Program Evaluation

Another use of birth defects surveillance data is program evaluation. Typically, this use is employed subsequent to research efforts, many of which were also based on surveillance data and may represent a baseline from which post-intervention improvement can be measured. Program evaluation is a valuable and desired area of activity with important scientific, academic, social, and policy applications. Program evaluation can focus on different aspects of surveillance program activities, such as case referrals and clinical interventions. First, evaluating a program for service referral can give investigators information on the efficacy of their referral agencies or the appropriateness of the services offered. Second, evaluating clinical

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intervention studies allows researchers to assess both the effectiveness of the intervention and the validity of their clinical assumptions.

One study involving program evaluation of a clinical intervention using birth defects surveillance data was performed in Nuevo León, México (Martinez de Villarreal et al., 2002). The investigators assessed the effectiveness of a folic acid campaign in reducing the occurrence of neural tube defects. Investigators first developed a base rate for neural tube defects prior to administration of the folic acid and counseling services. An intervention was then initiated that included five mg of folic acid supplementation per week, as well as counseling and social services. After 28 months, the rates of neural tube defects were ascertained again. From the baseline in 1999 (95 cases of neural tube defects), neural tube defects declined by 50% in the next two years (59 cases in 2000, 55 cases in 2001).

This study illustrates the wide range of uses for birth defects surveillance data in evaluation. First, data were used to assess an initial rate of neural tube defects and at the conclusion of the intervention to assess its appropriateness and efficacy. In addition, the study demonstrated the efficacy of folic acid supplementation in reducing the occurrence of neural tube defects and the fact that the methods of administration were clinically appropriate and effective.

In another example of the use of surveillance data in program evaluation, Meyer and Oakley (2000) used data from the North Carolina Birth Defects Monitoring Program to assess the folic acid fortification mandates of the federal government. The results suggested that the decline in the occurrence of neural tube defects was marginal and not the predicted 50% decrease. The authors' recommendation was to increase the folic acid fortification standards on a national level.

1.4.7 Clinical Research

Recently a group of researchers in the United Kingdom carried out surveillance in one Health Region using multiple sources to identify all individuals with specific conditions (Holland et al., 1998; Whittington et al., 2001). The condition the researchers captured that is of greatest relevance to birth defects is Prader-Willi syndrome (PWS). The first step was to conduct population-based surveillance in the Cambridge Health District (eight English counties with a base population of 280,000 individuals) (Whittington et al., 2001). The birth prevalence of PWS was estimated to be 1:22,000 and the mortality rate more than 3% per year. The next step was to carry out population-based clinical research about phenotypic features, including the prevalence of behavioral and health problems in PWS. Clarke et al. (2002) reported the prevalence of compulsive and similar behaviors among individuals with PWS in this population. Butler et al. (2002) presented data on the prevalence of comorbidities in PWS that could contribute to reduced life expectancy for persons with this condition. Most recently Holland et al. (2003) reported on the specific behaviors that comprise the proposed behavioral phenotype in PWS.

Although this work represents a non-traditional method of surveillance compared to state surveillance programs in the United States, it is important in terms of clinical research that has been conducted and the potential for conducting similar work using state-based surveillance data. A major advantage of these clinical studies is that they are population based. Even though all individuals identified through the surveillance work did not participate in the collection of behavioral and health data, the sample of individuals with PWS who participated in the clinical research can be compared to the total population of ascertained individuals to evaluate how representative the sample is of individuals in the Health Region who have PWS. Usually this is not possible using common methods of clinical research.

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1.4.8 Using Birth Defects Data in the Future

For the data collection process itself, abstracting methods continue to be refined. Quality assurance procedures and ongoing training, aimed at increasing data accuracy and validity, are being implemented in order to assure a certain level of fidelity and trust in the data collected. Improving and standardizing these procedures are among the objectives of these guidelines.

The future uses of birth defects surveillance data are related to scientific advances in other areas of research. Several developing scientific fields will utilize birth defects data in novel ways. For example, our understanding of molecular biology has developed exponentially. With the successful sequencing of the human genome, the resulting information will provide significant information on genetic factors influencing disease risk. Consequently, these discoveries will be investigated for certain genetic regulatory mechanisms and environmental triggers. Using birth defects surveillance data, investigators will be able to examine possible environmental exposures that are etiologically associated with birth defects in the presence of a particular genetic background. Discoveries of gene-environment interaction will allow researchers to understand etiologic associations. Additionally, the way in which these environmental conditions regulate gene expression will further illuminate these associations.

Future advancements in research supported by birth defects data will benefit from the integration of electronic medical records. Current methods for obtaining birth defects data are laborious. They frequently involve extensive abstraction procedures, reporting cases to the respective health department, entering the abstracts into the database, and categorizing the data. These methods will be streamlined, as medical records and birth defects surveillance systems are maintained electronically. This will have two general effects: first, it will help facilitate the abstraction process by eliminating bulky charts containing information not necessarily applicable to the birth defects surveillance program and, second, it will allow researchers to access these information-rich databases more quickly and efficiently. Furthermore, database search functions will allow researchers to identify cases of interest instantaneously without physically sifting through thousands of reported cases. Ultimately, researchers will be given access to the electronic surveillance database. Using surveillance systems researchers will able to search for cases of interest and refine their cohort by filtering cases by demographics, location, or maternal prenatal behaviors. A study that currently takes weeks to conclude would be completed in the course of several hours.

Researchers continuously find new and exciting uses of the data from birth defects surveillance programs. Given the breakthroughs achieved through earlier studies using surveillance data, the possibilities of future revelations are staggering. In their relatively short existence, birth defects surveillance programs have changed the ways in which professionals view birth defects both clinically and socially. The importance of the impact of birth defects surveillance programs on clinical and public health research cannot be overstated, as such research is revolutionizing the way scientists, clinicians, and health care professionals approach, treat, and manage infants affected by birth defects, while also advancing our understanding of preventive measures.

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Chapter 2 Legislation

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Acronyms

CDC Centers for Disease Control and Prevention

CE Covered Entity

FERPA Family Educational Rights and Privacy Act

DHHS Department of Health and Human Services

HIPAA Health Insurance Portability and Accountability Act

OCR Office of Civil Rights

PHA Public Health Authority

PHI Protected Health Information

PR Privacy Rule

Chapter 2 Legislation

2.1 Introduction

Legislation supporting birth defects surveillance activities is important for several reasons. For example, legislation serves to define the purposes of surveillance activities, specifies the kinds of data or information to be collected, and designates who is responsible for this activity. The first birth defects legislation was passed in New Jersey in 1926. During the past 20 years, the majority of states have enacted legislation mandating reporting of birth defects to the health department. As of April 2004, 41 states had existing legislation or rule related to birth defects surveillance.

Although there are a number of advantages to having legislation that supports birth defects surveillance, some limitations may also accrue. Early in their planning process, new or relatively new state programs should consider both the benefits and the possible limitations of birth defects surveillance legislation. At this early stage in a program's development, the opportunity exists to advocate for and perhaps assist in crafting clearly written, effective legislation that will serve the needs of the program in years to come.

In this chapter we discuss the distinction between the terms 'legislation', 'regulation', and 'authority' (Section 2.2); key elements of model legislation (Section 2.3); and federal laws that can affect birth defects surveillance programs (Section 2.4). References cited in this chapter may be found in Section 2.5.

To assist those interested in drafting or revising state legislation concerning birth defects surveillance, we append sample legislation from Arkansas, California, New Jersey, New York, Oklahoma, and Texas (see Appendix 2.1). Additional appendices include a table of birth defects legislation (Appendix 2.2), definitions used to determine 'covered entity' status under the Privacy Rule (Appendix 2.3), and an excerpt from the text of the Health Insurance Portability and Accountability Act (Appendix 2.4).

2.2 Legislation, Regulation, and Authority

'Authority' to mandate the reporting of birth defects to a surveillance program can be granted through 'legislation' or 'regulation'. In this section we explore distinctions among these and other related terms.

Legislation is the process of enacting laws by a legislative body. The type of law depends on the legislative authority granted. State legislatures and Congress have complex processes to enact legislation. These processes vary from state to state. In the simplest terms, state and federal legislative bodies create **statutory law**, also called a **legislative act**. These terms denote a bill that has been passed by one house in a bicameral legislature. After enactment by both houses, the terms 'law' and 'act' may be used interchangeably. A **statute** is the formal written enactment of a legislative body, whether federal, state, city, or county.

State and federal agencies are arms of the executive branch of the government. Such agencies have broad power granted under state and federal law to make *regulations* that govern activities for which they are responsible. Leaders of public health and other state agencies are not elected, but rather appointed by the executive, usually the governor of a state. Under current public health legislation, public health authorities may make regulations that can be mandatory, voluntary, directive, or prohibitive.

In sum, the term 'legislation' refers to a law enacted by an elected body, whereas 'regulations' are created by agencies.

For an agency, such as a state public health department, to establish a regulation mandating the reporting of birth defects, the health department must have the power or the *authority* to establish that type of regulation. This power can be based on state law or on an act of the executive power of the state, such as the governor. If the health department does not already have such regulatory power, then two options exist, namely, proposing a state law mandating birth defects reporting or proposing a state law granting authority to the health department to establish a regulation.

A state reporting law is straightforward and more democratic because it is enacted by elected representatives and gives an agency clear power or authority to do whatever the law states. However, a state reporting law also places the power to modify or change the law in the hands of the legislative body, despite the fact that the legislature may not be well informed about public health matters. Because most legislative bodies recognize the expertise of the people who run public health agencies, they generally grant them the necessary authority to conduct their work properly. Thus, the legislative bodies of many states have given the health department power to enact the regulations they deem necessary to protect the public health and welfare.

2.3 Key Elements of Model Legislation

Birth defects legislation should be considered early in the developmental phase of a surveillance program, if possible. This allows for legislation to be written clearly to support facilitation of surveillance activities. Language should be broad and flexible enough to cover all of the areas necessary to meet programmatic objectives, yet not to be so vague as to be confusing or meaningless. Well-written legislation that facilitates birth defects surveillance should address the key elements outlined in the Sections 2.3.1 through 2.3.8 below. These include:

- > Designation of agency authority
- > Purpose and priorities
- > Access to data and records
- Ability to share data while maintaining confidentiality
- > Terminology and definitions
- > Opt-out clauses
- > Advisory committee
- > Funding

2.3.1 Designation of Agency Authority

Model state legislation for birth defects surveillance should specify the agency that has the overall grant of authority for the system. This authority usually resides within the department of health, which has the power to enact rules and regulations, establish criteria for reportable conditions, and implement and oversee procedures for reporting. In most cases, there is no need to detail the specific regulations in the legislation. However, legislation should specify that the department has the authority to enact and enforce the regulations.

2.3.2 Purpose and Priorities

The purpose of the program will drive decision-making about its scope and activities. The purpose will also help states define outcomes, ages to be covered, and the most important sources of data to be included. Language should clearly articulate what the system should do and what its priorities should be. For example, Hawaii's legislation contains the following language:

"The department of health shall establish the statewide birth defects program to:

- 1) Collect surveillance information on birth defects and other adverse reproductive outcomes;
- 2) Report the incidence, trends and causes of birth defects and other adverse reproductive outcomes;
- 3) Report information for the development of prevention strategies to reduce the incidence of birth defects and other adverse reproductive outcomes; and
- 4) Develop strategies to improve the access of children with birth defects to health and early intervention services." (Hawaii Revised Statutes, Chapter 321, §321)

2.3.3 Access to Data and Records

Legislation should grant the birth defects surveillance program the authority to access hospital discharge data and medical records or to require reporting with access for follow-up as needed. Legislation that provides for access to medical records grants surveillance programs the opportunity to obtain more complete and reliable reporting of birth defects, while also ensuring that surveillance data sets are large enough to be useful to researchers and service providers.

California's birth defects surveillance law states that:

"... The director shall require health facilities, with 15 days' notice, to make available to authorized program staff the medical records of children suspected or diagnosed as having birth defects, including the medical records of their mothers. In addition, health facilities shall make available the medical records of mothers suspected or diagnosed with stillbirths or miscarriages and other records of persons who may serve as controls for interview studies about the causes of birth defects ..." (California Health and Safety Code, Part 2, Chapter 1, §103830)

Legislation with mandated reporting should include language that allows a program to access medical records for follow-up to ensure data quality. For example, New Jersey's legislation stipulates that:

"The Commissioner of Health, in consultation with the Public Health Council, shall require the confidential reporting to the Department of Health of all cases ..." (New Jersey, Chapter 26:8-40.2)

Then, in its regulations, the department of health addresses the follow-up component:

"Every health facility and independent clinical laboratory shall allow access to, or provide necessary information on infants with birth defects ..." (New Jersey Rules, Chapter 20, Subchapter 1, 8:20-1.2j)

2.3.4 Ability to Share Data While Maintaining Confidentiality

Legislation should specify who can have access to the data and how the confidentiality of the data will be protected. Many states have specific guidelines regarding the use of data for research purposes, and legislation may stipulate that persons who violate rules about data use or confidentiality are subject to civil penalties. For example, Texas' legislation states that:

- "(a) Access to the central registry information is limited to authorized department employees and other persons with a valid scientific interest who are engaged in demographic, epidemiological, or other studies related to health and who agree in writing to maintain confidentiality.
- (b) The department shall maintain a listing of each person who is given access to the information in the central registry. The listing shall include:
 - (1) the name of the person authorizing access;
 - (2) the name, title, and organizational affiliation of each person given access;
 - (3) the dates of access; and
 - (4) the specific purpose for which the information was used.

(c) The listing is public information, is open to the public under the open records law, Chapter 424, Acts of the 63rd legislature ... and may be inspected during the department's normal hours of operation." (Texas Health and Safety Code, Subchapter D, § 87.062)

2.3.5 Terminology and Definitions

Terminology should be defined clearly, but not in an overly narrow or restrictive manner. For instance, it is more effective to specify surveillance for the general category of 'birth defects' rather than for a narrow or finite list of specific defects such as spina bifida, anencephaly, Down syndrome, and so on.

The state of California defines birth defect as:

"... any medical problem of organ structure, function, or chemistry of possible genetic or prenatal origin." (California Health and Safety Code, Chapter 1, §103825 [a])

The legislation also specifies that *health facilities* are:

"... general acute care hospitals, and physician-owned or operated in clinics ... that regularly provide services for the diagnosis or treatment of birth defects, genetic counseling, or prenatal diagnostic services." (California Health and Safety Code, Chapter 1 §103830)

Broader language is more flexible, inclusive, and comprehensive than narrow language and allows for future modifications in program priorities or activities, whereas revising or amending narrowly written legislation can be a lengthy and difficult process. Legislating surveillance of specific defects may prove to be problematic in the long run as conditions change or as it becomes necessary or desirable to collect data on additional defects or combinations of defects. Definitions should be in the agency's regulations, not in the enabling legislation.

2.3.6 Opt-out Clauses

In most cases, parental consent is not required in order for a surveillance program to be able to collect data on children with birth defects from schools or health care providers. Some states, however, do require written consent from parents. Because obtaining written consent from parents can be problematic, some states handle this issue with an opt-out clause.

For example, Ohio's opt-out clause states that the health department shall adopt rules that will:

"Establish a form for use by parents or legal guardians who seek to have information regarding their children removed from the system and a method of distributing the form to local health departments ... and to physicians. The method of distribution must include making the form available on the internet." (Ohio, House Bill No.534, § 3705.35[e])

Opt-out clauses assume consent unless otherwise stated, allowing the surveillance program to collect data unless a child's parent or legal guardian submits a written request that their child's information be removed from the surveillance system. Opt-out clauses eliminate the need for providers and surveillance program staff to obtain written consent from parents and contribute to more complete data collection.

2.3.7 Advisory Committee

States that consider the potential impact of legislation in the planning stages of their programs have the advantage of influencing the development of legislation that can support the overall growth and development of the program. In some states, for example, legislation calls for establishing an advisory committee to provide guidance and oversight for the design and implementation of birth defects surveillance. Advisory committees made up of experts from fields such as epidemiology, hospital administration, biostatistics, maternal and child health, and public health can develop recommendations and provide the expertise necessary to ensure that the program meets well-defined standards and goals. Some advisory committees also include parents of children with birth defects. For example, Vermont's legislation calls for the establishment of a 'birth information council'.

- "(a) The commissioner of health, in collaboration with the March of Dimes, shall appoint a birth information council to advise on the need for and implementation of a comprehensive, integrated, and confidential birth information system.
- (b) The council shall be composed of nine members, who represent each of the following interests:
 - (1) obstetrics and gynecology;
 - (2) pediatrics and genetics;
 - (3) the Vermont Children's Health Improvement Program;
 - (4) a parent of a child with special medical needs;
 - (5) an adult with special medical needs;
 - (6) the commissioner of health, or his or her designee;
 - (7) the Family, Infant, and Toddler Program;
 - (8) the Vermont chapter of the March of Dimes; and
 - (9) the Vermont Program for Quality Health Care." (Vermont, H.636, § 5084)

2.3.8 Funding

Cost can be an impediment to establishing a birth defects surveillance system.

Some states have legislation mandating special funds to cover the operating expenses of their birth defects surveillance program. Sources of special funds include marriage license, birth certificate, and newborn screening fees. For example, Iowa's special fund is supported through birth registration fees:

"It is the intent of the general assembly that the funds generated from the registration fees be appropriated and used as follows:

- (1) Beginning July 1, 2003, and ending June 30, 2005 ... five dollars of each fee for the birth defects institute central registry established pursuant to section 136A.6.
- (2) Beginning July 1, 2005, ... ten dollars of each fee for the birth defects institute central registry established pursuant to section 136A.6." (Iowa Code, §144.13A)

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In summary, paying due consideration to how legislative language can affect the design, implementation, and operation of the surveillance program and further ensuring that the birth defects surveillance program itself has input into legislative language from the time the program is established can have a significant impact on the long-term success of the program.

Chapter 2 2-7 Legislation

2.4 Federal Laws

A broad range of federal laws must be considered when planning state legislation, local regulations, or new birth defects surveillance programs. While state laws will govern most of the activities of the program, the impact of federal privacy regulations must also be considered. Depending upon how the birth defects program is structured, it may need to follow the Health Insurance Portability and Accountability Act (HIPAA) discussed in Section 2.4.1, the Family Educational Rights and Privacy Act (FERPA) discussed in Section 2.4.2, and other federal regulations such as the Privacy Act (Section 2.4.3), the Public Health Service Act (Section 2.4.4), and the Freedom of Information Act (Section 2.4.5). The following sections provide basic information about major federal laws that must be considered when setting up a birth defects surveillance program. In Section 2.4.6 we discuss the supportive role that can be played by state health officials or staff of the Centers for Disease Control and Prevention (CDC) in conjunction with planning state legislation or local regulations for birth defects surveillance programs.

2.4.1 Health Insurance Portability and Accountability Act (HIPAA)

The Health Insurance Portability and Accountability Act was passed in 1996 to protect consumers of the insurance industry. The Privacy Rule (or PR, also referred to as the Rule), which implements the Act, became effective on April 14, 2001, and creates national standards to protect an individual's medical records and other personal health information, known as *protected health information* (or PHI). The Rule gives patients more control over their health information and establishes appropriate safeguards that health care providers and other *covered entities* (or CEs) must establish to protect the privacy of PHI. Violators are subject to civil and criminal penalties if they violate patients' privacy rights as stated in the Privacy Rule. The Rule allows for disclosure of some forms of data for activities carried out by *public health authorities* (or PHAs) but limits release of information to the minimum necessary for the purpose of the disclosure. In addition, the covered entity may rely on the public health authority for what constitutes the 'minimum necessary'.

The Privacy Rule requires health care providers who are covered entities to provide information to patients about their privacy rights and how their information can be used, to adopt clear privacy procedures and adequately train employees in these procedures, and to designate an individual to be responsible for seeing that the privacy procedures are adopted and followed. Privacy protections should not, however, interfere with a patient's access to health care or the quality of health care delivered.

Basic Provisions of the Privacy Rule That Affect Birth Defects Reporting

A state, county, or local health department that performs functions that make it a covered entity, or otherwise meets the definition of a covered entity, may elect to call itself a *hybrid entity*. For example, a state Medicaid program is a covered entity (i.e., a health plan) as defined in the Privacy Rule. Some health departments operate health care clinics and thus are health care providers. If these health care providers transmit health information electronically, in connection with a transaction covered in the HIPAA Transactions Rule, they are covered entities.

Most of the requirements of the Privacy Rule apply only to the hybrid entity's health care provider component(s). If a health department elects to be a hybrid entity, there are restrictions on how its health care component(s) may disclose protected health information to other components of the health department. Birth defects surveillance components that provide genetic counseling and other types of

health care services will most likely be required to comply with the Rule's 'covered entities' provisions, if they bill electronically for their services. (See 45 C.F.R. § 164.504 (a) – (c) for more information about hybrid entities.)

For further information, see the definitions of 'covered entity', 'health care provider', 'health plan', and 'health care clearinghouse' in 45 C.F.R. §160.103. See also, the "Covered Entity Decision Tools" posted at:

http://www.cms.gov/hipaa/hipaa2/support/tools/decisionsupport

Uses and Disclosures for Which an Authorization or Opportunity to Agree or Object Is Not Required

Section 164.512 of the Privacy Rule sets forth the conditions under which a covered entity, as defined previously, may disclose protected health information without the individual's consent or authorization. Below is a discussion of the application of the Rule to the birth defects surveillance system. The actual text of the regulation can be found in Appendix 2.4.

Consent and notice. The US Department of Health and Human Services (DHHS) made changes to the Privacy Rule effective August 14, 2002, to protect privacy while eliminating barriers to treatment. The notice requirement was strengthened, making consent for routine health care delivery purposes optional. The Rule requires covered entities to provide patients with notice of a patient's privacy rights and the privacy practices of the covered entity. The strengthened notice requires direct treatment providers to make a good faith effort to obtain patients' written acknowledgement of the notice of privacy rights and practices. The modified Rule removes mandatory consent requirements while providing covered entities with the option of developing a consent process that works for that entity. The Rule also allows consent requirements already in place to continue, but does not mandate any particular standard.

In states where data collection for birth defects surveillance is ongoing and there is no mandatory reporting law, it would be helpful to approach the data source with a request to have the public health authority listed in the privacy notice that is provided to patients. Note, however, that this does not circumvent the accounting provisions of the Rule for the covered entity.

Mandatory reporting – 'Required by law' versus 'permitted'. Extensive discussion has ensued within the public and private health care sectors regarding the need for mandatory reporting laws in states in order for birth defect surveillance programs to collect data. Note that this section of the Rule, §164.512, has two subsections.

- (a) Standard: uses and disclosures required by law.
- (b) Standard: uses and disclosures for public health activities.

Subsection (a) is the provision for disclosures that are required by law. If a state has a mandatory birth defects reporting law, then this is the provision in the Privacy Rule that allows that law to remain intact. The definitions in the section below explain what 'required by law' means under the Privacy Rule. However, if a state health department meets the definition below of a public health authority, then the

health department may have authority to collect birth defects data based on the department's broad grant of authority from the state to protect and promote health, prevent and control disease, or other activity.

As noted earlier, each state health department has specific authority granted it under the laws of that state. Most health departments do have some regulatory authority and can, therefore, make birth defects reporting mandatory under that authority. If the health department does not have the present authority to make such a regulation, or conduct such activity, then the health department may request that this authority be granted by the legislature, after which the department may promulgate its regulation. This method is acceptable under the Privacy Rule.

The most significant distinction to make is that subsection (a) is for reporting *required by law*, whereas subsection (b) is for reporting *authorized by law*. Although there is no definition of 'authorized by law' in the Rule, DHHS has sought to make this point more clearly in the Preamble to the Rule (64 FR, page 59929):

"When we describe an activity as 'authorized by law,' we mean that a legal basis exists for the activity. The phrase 'authorized by law' is a term of art that includes both actions that are permitted and actions that are required by law."

In addition to this comment, new Office of Civil Rights (OCR) guidelines state:

"The HIPAA Privacy Rule permits disclosures that are required by law. Furthermore, disclosures to public health authorities that are authorized by law to collect or receive information for public health purposes are also permissible under the Privacy Rule." (OCR HIPAA Privacy Dec 3, 2002, http://www.hhs.gov/ocr/hipaa)

In short, public health authorities have two different paths by which to access data for surveillance, a mandatory reporting law, or the regulatory or program authority to collect the data. (See Appendix 2.4 for OCR HIPAA privacy regulation text.)

Data Sharing and Public Health Authorities

A public health authority that has either a mandatory reporting law, or a regulation, or some other grant of authority to collect data under the previously discussed §164.512, may use those data in any way that is permitted under state and federal law. Data that are collected by a third party, such as a university, under a grant or a contract on behalf of a public health authority, such as the Centers for Disease Control and Prevention (CDC), whether a bona fide agent or not of that health department, falls under the Privacy Rule definition of a 'public health authority':

"'Public health authority' means an agency or authority of the United States, a State, a territory, a political subdivision of a State or territory, or an Indian tribe, or a person or entity acting under a grant of authority from or contract with such public agency, including the employees or agents of such public agency or its contractors or persons or entities to whom it has granted authority, that is responsible for public health matters as part of its official mandate." $(45 \ CFR \ \$164.512(b)(l)(i))$

The Rule does not comment on what the public health authority may or may not do with the data it has legally collected. HIPAA seeks to regulate the release and use of protected health information by covered entities, and a public health authority is not a covered entity under the Rule (unless they have designated themselves as such). The grantee, holder of a cooperative agreement, or contractor conducting a public health activity, as a public health authority, as defined above, may share the data in ways that comport with all previously promulgated laws and regulations. Once data are in the possession of a public health

authority, the Rule should not be an issue for the PHA because the Rule does not regulate the use or disclosure of protected health information by a PHA.

A number of health departments have designated some of their components as covered components because they provide health care as defined in the Rule. In this case, the entire health department may be called a 'hybrid entity'. The consequences for data sharing are the same as if the designated component, or covered entity, were any other health care provider. The covered entity component of the health department can share the data it collects from individuals with the non-covered PHA component of the health department. The covered entity would have to provide the individual with the 'notice of privacy practices', which would include information to the effect that the covered entity was sharing data with other components of the health department. The covered component would also have to comply with all other provisions of the Rule, including accounting for disclosures to public health authorities. Some health departments may even provide consents to the individual based on the requirements of a state or local requirement, or to increase public confidence in the health department.

Nor is the data-sharing that flows from a public health authority to a covered entity after data collection regulated by the Privacy Rule. In cases where the public health authority wishes to refer a case to another covered entity, such as a health care provider, for a public health intervention, and the covered entity may report back its findings, remember that the definition of 'public health activities' includes the following:

"A public health authority that is authorized by law to collect or receive such information for the purpose of preventing or controlling disease, injury, or disability, including, but not limited to, the reporting of disease, injury, vital events such as birth or death, and the conduct of public health surveillance, public health investigations, and public health interventions." (45 CFR \$164512(b)(l)(i))

When requesting data from a covered entity, it is also important to note that even though public health authorities are exempted from the need for the authorization of the person for disclosure, the covered entity is only required to provide for the minimum necessary information to accomplish the public health mission of the PHA. In addition, the covered entity may, under the Rule, reasonably rely on the representation of the PHA for what constitutes the 'minimum necessary' information.

Some state grantees conducting birth defects and other kinds of surveillance funded by CDC have asked what kind of proof of identification (ID) they need to show to the covered entity to assure them that they are in fact a PHA and have the authority to obtain the data they seek from the CE. Business cards, government identification badges, letterhead, or other types of official representation are sufficient. Because there are so many different types of ID, DHHS chose to be very broad in this area by not specifying one type.

Data Clearinghouses and Business Associates

Some state health departments do not carry out actual surveillance and data collection; instead, hospitals voluntarily report birth defects data to a data collection entity or clearinghouse that compiles the data and then reports the information in some form to the health department. In these cases, the hospital and clearinghouse are required to execute a data use agreement, and the covered entity must disclose this information in the privacy notice provided to patients. The clearinghouse may provide the data to the public health authority under that Rule just as the covered entity could do, without the authorization or consent of the person for purposes of public health activities, surveillance, and, under some circumstances, research.

Surveillance versus research under the Privacy Rule. Research is covered under a separate section of the Privacy Rule. Unlike the public health authority provisions discussed above, the research provisions do not exempt public health authorities from compliance with the Rule as research is not a public health activity as defined in the Privacy Rule. The Rule defines *research* as:

"A systematic investigation, including research development, testing, evaluation, designed to develop or contribute to generalizable knowledge." (45CFR 164.501)

The recent revision in the Privacy Rule sought to bring the definition of 'research' in the Rule in line with the definition for the same term in the Common Rule. The Common Rule definition of 'research' is the one used by CDC (45 CFR 46.102[e]).

De-identified data use. For research purposes, a covered entity may always use or disclose health information that has been de-identified (45 CFR 164.502(d) and 164.514[a]-[c]). The Rule has a very strict definition of 'de-identified' that truly eliminates all possibility of re-identification of the individual. However, a covered entity may enter into a data use agreement with a researcher that would allow the CE to disclose to the researcher a limited data set for the purposes of research, public health, or health care operations (45 CFR 164.514[e]). A *limited data set* is specifically defined in the Privacy Rule to exclude certain direct identifiers; however, the limited data set contains sufficient geographical and vital information – such as birth, death, admit and discharge data – that it can be very useful for birth defects research. In addition, there are other specific requirements that must be included in the data use agreement. These include:

- > Stating the permitted uses and disclosures of the limited data set
- > Limiting who can receive the data
- > Requiring the researcher to agree to:
 - Abide by and not violate a data use agreement
 - Protect the data from re-disclosure
 - Report any unauthorized use or disclosure
- ➤ Binding all contractors or agents to the data use agreement
- > Refraining from identifying or contacting the individual

Another way to obtain access to protected health information for research without authorization from the individual is to obtain documented Institutional Review Board (IRB) or Privacy Board approval for an exemption (45 CFR 164.512[i][l][i]). This provision is most practical for conducting records searches when use of de-identified data is not useful. There are extensive requirements under this section of the Rule that must be adhered to. Another way to obtain access to data for research without authorization of the individual is when preparing a research protocol preparatory to research (45 CFR 164.512 [i][l][ii]). Except for these limited exceptions, the disclosure or use of protected health information for research purposes requires the written authorization of the individual.

2.4.2 Family Educational Rights and Privacy Act (FERPA)

The Family Educational Rights and Privacy Act (20 U.S.C. § 1232g; 34 CFR Part 99) is a federal law that protects the privacy of student education records. The law applies to all schools that receive funds under an applicable program of the US Department of Education. There are some privately funded schools to which FERPA does not apply.

FERPA gives parents specific rights with respect to their children's educational records. These rights transfer to the student when he or she reaches the age of 18 or attends a school beyond the high school level. Students to whom the rights have transferred are defined as *eligible students* in FERPA.

- Parents or eligible students have the right to inspect and review the student's education records maintained by the school.
- > Parents or eligible students have the right to request that a school correct records that they believe to be inaccurate or misleading.
- ➤ Generally, schools must have written permission from the parent or eligible student in order to release any information from a student's education record.

However, FERPA allows schools to disclose those records, without consent, to the following parties or under the following conditions (34 CFR § 99.31):

- > School officials with legitimate educational interest
- > Other schools to which a student is transferring
- > Specified officials for audit or evaluation purposes
- Appropriate parties in connection with financial aid to a student
- Organizations conducting certain studies for or on behalf of the school
- > Accrediting organizations
- Appropriate officials in cases of health and safety emergencies
- > State and local authorities, within a juvenile justice system, pursuant to specific state law
- To comply with a judicial order or lawfully issued subpoena

Access to educational records can be necessary to a birth defects surveillance program for follow-up and early intervention services. FERPA generally prohibits access to educational records without the prior written consent of the parent or guardian.

Surveillance versus research under FERPA. For compliance with FERPA, there is no distinction made between surveillance and research. The issue in FERPA is who holds the data and who wants access to the data and why. The fact that the information in the educational record is medical, behavioral, sociological, or psychological in nature in no way alters the inability to access the information without parental consent. All information, other than student directory information, in an educational record maintained by a school, regardless of the nature of the information, is considered to be an educational record. It is important to note that HIPAA specifically states that nothing in HIPAA in any way alters FERPA. As a result, FERPA, unlike HIPAA, defines its 'protected records' simply by who possesses them, whereas in HIPAA the analysis of what is protected and the exceptions are more complex.

2.4.3 Privacy Act

The Privacy Act of 1974, 5 U.S.C. § 552a (2000), which has been in effect since September 27, 1975, can generally be characterized as an omnibus 'code of fair information practices' that attempts to regulate the collection, maintenance, use, and dissemination of personal information by federal executive branch agencies. However, the Act's imprecise language, limited legislative history, and somewhat outdated regulatory guidelines have rendered it a difficult statute to decipher and apply. Moreover, even after more than 25 years of administrative and judicial analysis, numerous Privacy Act issues remain unresolved or unexplored. Adding to difficulties in interpretation is the fact that many Privacy Act cases are unpublished district court decisions. The general rule contained in the Privacy Act is:

"No agency shall disclose any record which is contained in a system of records by any means of communication to any person, or to another agency, except pursuant to a written request by, or with the prior written consent of, the individual to whom the record pertains [subject to 12 exceptions]." (5 U.S.C. § 552a[b])

States have adopted similar laws that should be considered when drafting legislation for a birth defects surveillance program. For further information, see the Department of Justice website at http://www.doj.gov.

2.4.4 Public Health Service Act

The Public Health Service Act of July 1, 1944 (42 U.S.C. §201), consolidated and substantially revised all existing legislation relating to the US Public Health Service, of which the CDC is a part. The Public Health Service Act is a broad compilation of authorities under which CDC administers national and international programs for the prevention and control of communicable and vector-borne diseases and other preventable conditions. The Public Health Service Act is only applicable to federal agencies within the Public Health Service.

Title III of the Public Health Service Act sets forth the general powers and duties of the Public Health Service. Within this title, Sections 301, 307, 311, and 317 provide CDC and other agencies within the Service with general operating authorities, including but not limited to:

- Encourage, cooperate with and render assistance to other appropriate public health authorities, scientific institutions, and scientists in the conduct and promotion of activities relating to the causes, diagnosis, treatment, control, and prevention of physical and mental diseases.
- Make grants-in-aid to universities, hospitals, laboratories, and other public and private research institutions.
- ➤ Participate with other countries in cooperative endeavors in biomedical research, health care technology, and health services research for the purpose of advancing the status of health sciences in the United States.
- Cooperate with and assist states and their political subdivisions in the prevention and suppression of communicable diseases and other public health matters.

In regard to provisions of the Public Health Service Act which promote, encourage, and influence activities in the area of birth defects study and prevention, Section 317C was added to the Public Health Service Act by the Children's Health Act of 2000. Section 317C provides the general operating authority for the National Center on Birth Defects and Developmental Disabilities (NCBDDD), a center within the

CDC. This authority was recently renewed in accordance with the Birth Defects and Developmental Disabilities Prevention Act of 2003. In part, Section 317C allows NCBDDD to:

- ➤ Collect, analyze, and make available data on birth defects and developmental disabilities.
- Operate regional centers for the conduct of applied epidemiological research on the prevention of such defects and disabilities.
- > Provide information and education to the public on the prevention of such defects and disabilities.

The Public Health Service Act is codified in Title 42 of the United States Code.

2.4.5 Freedom of Information Act 5 USC §522 (FOIA)

All federal agencies are generally required under the Freedom of Information Act (FOIA) to disclose records they maintain when requested in writing by any person. Most states have adopted state laws that mirror the federal law. Therefore, it is important for a state birth defects surveillance program to be aware of the state law and know which records they may have to provide to the public when requested. However, federal agencies may withhold information pursuant to nine exemptions and three exclusions contained in the statute, and states have generally adopted similar exemptions. The exemptions that are most pertinent here are the FOIA exemptions 3 and 6.

Exemption Number 3:

Specifically exempted from mandatory disclosure by statute (other than the Privacy Act), provided that such statute:

- (i) Requires that the matters be withheld from the public in such a manner as not to leave any discretion on the issue, or
- (ii) Establishes particular criteria for withholding or refers to particular criteria for withholding or refers to particular types of matters to be withheld.

This exemption is useful for protecting birth records in surveillance programs when the authorizing legislation specifically exempts the information in the statute.

Exemption Number 6:

Personnel and medical files and similar files the disclosure of which would constitute a clearly unwarranted invasion of personal privacy.

The FOIA applies only to federal agencies and does not create a right of access to records held by Congress, the courts, or by state or local government agencies. Each state has its own public access laws that should be consulted for access to state and local records. Each federal agency is responsible for meeting its FOIA responsibilities for its own records. Likewise, each federal agency component is responsible for processing FOIA requests for the records that it maintains. For more information and a list of FOIA federal contacts, see the Department of Justice website at http://www.doj.gov.

2.4.6 Advocacy

In this section we discuss advocacy for the development and implementation of surveillance systems in terms of both the state's role and CDC's role in such advocacy.

The role of the state in advocacy. State health officials and surveillance staff can be important partners for advocates in the development and implementation of surveillance systems. While state employees may be limited in terms of what activities they can participate in within advocacy, they can work together with advocates throughout the process in order to create or improve birth defects systems. State officials and health department surveillance staff bring planning, technical assistance, and an understanding of the political environment to the planning and implementation process.

The role of the CDC in advocacy. The CDC can also work with states and with advocates to provide technical assistance in the design, planning, and implementation stages of a birth defects surveillance system and can make recommendations for improving ongoing programs. CDC can also play a substantial role in educating policymakers and the public about the benefits of a birth defects surveillance program.

2.5 References

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Appendix 2.1 Sample State Legislation

Appendix 2.1 – Sample State Legislation

Appendix 2.1.1 Arkansas Legislation	A2.1.1-1
Appendix 2.1.2 California Legislation	A2.1.2-1
Appendix 2.1.3 New Jersey Legislation	A2.1.3-1
Appendix 2.1.4 New York Legislation	A2.1.4-1
Appendix 2.1.5 Oklahoma Legislation	A2.1.5-
Appendix 2.1.6 Texas Legislation	A2.1.6-1

Appendix 2.1.1 Arkansas Legislation

Arkansas Code of 1987 Annotated Title 20. Public Health and Welfare Subtitle 2. Health and Safety Chapter 16. Reproductive Health Subchapter 2. Arkansas Reproductive Health Monitoring System

20-16-201 Establishment- Purpose

- a) The Arkansas Reproductive Health Monitoring System is established and is to be administered within the Arkansas Children's Hospital.
- b) The purpose of the system is to collect and analyze data from a number of sources to describe trends in the occurrence of reproductive endpoints such as congenital anomalies, fetal death, developmental disorders, etc., and to correlate those trends and investigate and report on the suspected causes of unexpected deviations in those trends

History. Acts 1985, No. 214, § 1; A.S.A. 1947, § 82-4608.

20-16-202 Definitions

As used in this subchapter, unless the context otherwise requires:

- 1) "Board" means the technical advisory board established in § 20-16-204;
- 2) "Commission" means the advisory commission established in § 20-16-203; and
- 3) "System" means the Arkansas Reproductive Health Monitoring System.

History. Acts 1985, No. 214, § 3; A.S.A. 1947, § 82-4610.

20-16-203 Advisory Commission- Members- Functions

- a) The Arkansas Reproductive Health Monitoring System shall be administered with the advise of an advisory commission appointed to one-year renewable terms by the Medical Director of the Arkansas Children's Hospital.
- b) The functions of the commission are to:
 - 1) Advise the medical director as to the adequacy of policies, procedures, and performance of the system;
 - Appoint members of the board upon the recommendations of the medical director;
 - Promote the purposes of the system and assist in identification of appropriate funding sources;
 - 4) Promote interagency cooperation toward the goals of this system;
 - 5) Advise the medical director regarding requests for data dissemination; and
 - Review mechanisms ensuring the maintenance of the confidentiality of personal data.
- c) This commission shall be composed of the following state agencies, professional members, and public members:
 - 1) The medical director of the Arkansas Children's Hospital;
 - 2) The chancellor of the University of Arkansas for Medical Science;
 - 3) The director of the Department of Health;
 - 4) The director of the Department of Human Services;
 - 5) The director of the Arkansas Department of Environmental Quality;
 - 6) The director of the National Center for Toxicological Research;
 - 7) One (1) representative of the Arkansas Medical Society;
 - 8) One (1) representative of the Arkansas Academy of Pediatrics;

- 9) One (1) representative of the Arkansas Society of Obstetrics & Gynecology;
- 10) One (1) representative of the Arkansas Hospital Association;
- 11) One (1) representative of the State Plant Board;
- 12) Two (2) consumer representatives;
- 13) One (1) member from the Senate Public Health, Welfare, and Labor Committee and one (1) member from the House Public Health, Welfare, and Labor Committee; and
- 14) Up to four (4) additional members at large may be appointed.
- d) Members of the commission who are not employees of the state may receive expense reimbursement in accordance with § 25-16-901 et seq.

History. Acts 1985, No 214, §§ 4, 11; A.S.A. 1947, §§ 82-4611; Acts 1997, No. 250, § 188; 1999, No. 1164, § 173.

Amendments. The 1997 amendment rewrote (d). The 1999 amendment substituted "Environmental Quality" for "Pollution Control and Ecology" in (c) (5).

20-16-204 Technical Advisory Board- Members- Functions

- a) There shall be a technical advisory board whose function shall be to:
- b) (1) This board shall be appointed to one-year renewable terms by the Medical Director of the Arkansas Children's Hospital upon recommendation of the commission and the director.
 - (2) It shall be comprised of a maximum of ten (10) regular members drawn from fields of expertise such as: medicine, industrial hygiene and toxicology, agriculture, environmental sciences, and epidemiology and statistics.
 - (3) At the discretion of the board and the director, ad hoc members of the board may be appointed for specific periods to advise on special needs or problems, which have been identified.
- c) Members of the board who are not employees of the state may receive expense reimbursement in accordance with § 25-16-901 et seq.

History. Acts 1985, No 214, §§ 5, 11; A.S.A. 1947, §§ 82-4612, 82-4618; Acts 1997, No 250, § 189.

Amendments. The 1997 amendment rewrote (c).

20-16-205 Director- Appointment- Power and duties

- a) The Arkansas Reproductive Health Monitoring System shall be administered by a director appointed by the Medical Director of the Arkansas Children's Hospital from among the professional staff of the Arkansas Children's Hospital.
- b) The director shall:
 - 1) Supervise the work of the system and administer the budget;
 - Appoint and remove such other employees as may be necessary to perform the duties and responsibilities of the system; and
 - 3) Select and retain the services of consultants whose advice is considered necessary to carry out the system's mandate.

History. Acts 1985, No 214, § 2; A.S.A. 1947, § 82-4609.

20-16-206 Authority to contract for information

- a) The Arkansas Reproductive Health Monitoring System is expressly authorized to contract for the production of any information, which its technical advisory board determines to be relevant to monitoring reproductive health from any department or agency of the state.
- b) Information shared under this section includes, but is not limited to, information identified by the name or other personal identifier, including information concerning any system by which such data or information is identified or classified if required to decipher the information.

History. Acts 1985, No 214, § 6; A.S.A. 1947, § 82-4613.

20-16-207 Information confidential- Exception

The Arkansas Reproductive Health Monitoring System is expressly exempted and prohibited from supplying any information by individual name or other personal identifier or in a form other than a statistical report or other appropriate form which protects the confidentiality of individuals except to any state agency or department which originally supplied the information to the system unless both the originating agency and the system grant release of this information for a specific purpose.

History. Acts 1985, No 214, § 7; A.S.A. 1947, § 82-4614.

20-16-208 Furnishing of information by hospitals

- All hospitals with patient records containing information pertaining to reproduction and development are required to share information in those records with the Arkansas Reproductive Health Monitoring System.
- b) No hospital shall be required to furnish information under this section until appropriate reimbursement in return for the service has been determined by the advisory commission and funds are available to pay the compensation.

History. Acts 1985, No 214, § 8; A.S.A. 1947, § 82-4615.

20-16-209 Furnishing of information by physician, clinic, etc.

- Any physician, clinic, person, or organization may provide information relative to reproductive health to the Arkansas Reproductive Health Monitoring System.
- b) No liability of any kind or character for damages or other relief shall arise or be enforced against any person or organization by reason of having provided the information or by reason of having released or published the findings of the system in order to reduce morbidity and mortality or to advance medical research or medical education.

History. Acts 1985, No 214, § 9; A.S.A. 1947, § 82-4616.

20-16-210 Intergovernmental agreements

The Arkansas Reproductive Health Monitoring System shall have the power to enter into agreements with neighboring states and the federal Centers for Disease Control and Prevention consistent with the requirements and restrictions of this subchapter in order to obtain relevant information for the system concerning Arkansas residents who receive health-related services outside the state.

History. Acts 1985, No 214, § 10; A.S.A. 1947, § 82-4617.

20-16-211 Funding and implementation

- a) The Arkansas Reproductive Health Monitoring System shall have the power to receive and expend grants, donations, and funds from public and private sources to carry out its responsibilities under this subchapter.
- b) The Arkansas Children's Hospital is not required to implement this system unless sufficient funds are available as determined by the Medical Director of the Arkansas Children's Hospital.
- c) The system may be implemented in stages or phases. **History.** Acts 1985, No 214, § 13; A.S.A. 1947, § 82-4620.

20-16-212 Reports

The Arkansas Reproductive Health Monitoring System shall periodically prepare reports of its findings for dissemination to appropriate agencies and interested persons.

History. Acts 1985, No 214 § 14; A.S.A. 1947, § 82-4621.

20-16-213 Rendering of patient care and regulatory activity prohibited

The Arkansas Reproductive Health Monitoring System is expressly prohibited from rendering patient care, promulgating any rule or regulation, or engaging in any regulatory activity.

History. Acts 1985, No 214, § 13; A.S.A. 1947, § 82-4620.

20-16-214 No actionable right, presumptions, or findings created

- a) Persons other than the state or Arkansas Reproductive Health Monitoring System shall not acquire any actionable right by virtue of this subchapter
- b) A determination by this system that a source is suspected of causing adverse reproductive health outcomes shall not create by reason thereof any presumption of law or finding of a fact which shall inure to, or be for, the benefit of any person other than the state.

History. Acts 1985, No 214, § 12; A.S.A. 1947, § 82-4619.

Arkansas Code of 1987 Annotated Title 20. Public Health and Welfare Subtitle 2. Health and Safety Chapter 16. Reproductive Health Subchapter 4. Reproductive Health Information

20-16-402 Information from state agencies

- a) (1) Any bona fide appropriately licensed medical facility, including, but not limited to, county hospitals, participating in recognized research in Arkansas and the federal Centers for Disease Control and Prevention are expressly authorized to contract for the production of any information relevant to monitoring reproductive health from any department or agency of the state.
 - (2) Information acquired under this subsection (a) includes, but is not limited to, information identified by name or other personal identifying information including the methods by which the information was compiled or tabulated.
- b) The University of Arkansas for Medical Sciences, Arkansas Children's Hospital, other participating medical facilities as described in subsection (a) of this section, and the federal Centers for Disease Control and Prevention are expressly prohibited from supplying any information obtained pursuant to subsection (a) of this section by individual name or other personal identifying information or in a form other than a statistical report or other appropriately form which protects the confidentiality of individuals.
- c) Information obtained pursuant to subsection (a) of this section may be returned to any state agency or department from which it was originally obtained. **History.** Acts 1983, No 773, §§ 1, 3; A.S.A. 1947, §§ 82-4601, 82-4603.

Appendix 2.1.2

California Legislation

CALIFORNIA BIRTH DEFECTS MONITORING PROGRAM

STATUTORY AUTHORITY SCIENTIFIC

FOUNDATIONS

SERIES

OCTOBER 1998

STATUTORY AUTHORITY

Recognizing that birth defects are a public health problem about which too little is known, the State Legislature in 1982 created the California Birth Defects Monitoring Program. From 1982-1990, seven pieces of legislation were passed and enacted, mandating the Program to:

- Maintain an ongoing birth defects monitoring program statewide
- Track birth defects rates and trends
- Evaluate whether environmental hazards are associated with birth defects
- Investigate other possible birth defects causes
- Develop birth defects prevention strategies
- Conduct interview studies about causes
- *Operate by contract with a qualified entity.*

This document includes the Program's current statutory authority in the Health & Safety Code.

HEALTH & SAFETY CODE, DIVISION 102, PART 2, CHAPTER 1

CHAPTER 1. BIRTH DEFECTS MONITORING PROGRAM

Section

103825.	Legislative findings and declaration.
103830.	Collection of information; system
	establishment; medical records.
103835.	Scope of program; assessment of resources
103840.	Investigative studies.
103845.	Advisory committee; membership.
103850.	Confidentiality of information; research;
	review and approval; civil penalty.
103855.	Contract for establishment and

implementation of program.

Chapter 1 was added by Stats.1995, c. 415 (S.B. 1360), § 4.

Historical and Statutory Notes Legislative findings relating to the nonsubstantive effect of Stats.1995, c. 415 (S.B. 1360), and the legislative intent not to create any new rights, see Historical and Statutory Notes under Health and Safety Code § 100100.

§ 103825. Legislative findings and declaration

The Legislature hereby finds and declares that birth defects, stillbirths, and miscarriages represent problems of public health importance about which too little is known; that these conditions lead to severe mental anguish on the part of parents and relatives and frequently to high medical care costs; and that a system to obtain more information about these conditions could result in development of preventive measures to decrease their incidence in the future. Therefore, it is the intent of the Legislature in enacting this section to accomplish all of the following:

- (a) To maintain an ongoing program of birth defects monitoring statewide. "Birth defect" as used in this chapter means any medical problem of organ structure, function, or chemistry of possible genetic or prenatal origin.
- (b) To provide information on the incidence, prevalence, and trends of birth defects, stillbirths, and miscarriages.
- (c) To provide information to determine whether environmental hazards are associated with birth defects, stillbirths, and miscarriages.

- (d) To provide information as to other possible causes of birth defects, stillbirths, and miscarriages.
- (e) To develop prevention strategies for reducing the incidence of birth defects, stillbirths, and miscarriages.
- (f) To conduct interview studies about the causes of birth defects
- (g) To affirm the authority of the state department to contract with a qualified entity to operate the birth defects monitoring program statewide.

(Added by Stats.1995, c. 415 (S.B., 1360), § 4.)

Historical and Statutory Notes

Derivation: Former §10800, added by Stats.1982, c. 204, § 1, amended by Stats.1985, c. 1137, § 1; Stats.1989, c. 8, § 1; Stats.1990, c. 122, § 1.

§ 103830. Collection of information; system establishment; medical records

The director shall maintain a system for the collection of information, necessary to accomplish the purposes of this chapter. The director shall require health facilities, with 15 days' notice, to make available to authorized program staff the medical records of children suspected or diagnosed as having birth defects, including the medical records of their mothers. In addition, health facilities shall make available the medical records of mothers suspected or diagnosed with stillbirths or miscarriages and other records of persons who may serve as controls for interview studies about the causes of birth defects. If it is necessary to photocopy records made available under this section, copying expenses shall be paid by the state department.

"Health facilities" as used in this section means general acute care hospitals, and physician-owned or operated clinics, as defined in Section 1200, that regularly provide services for the diagnosis or treatment of birth defects, genetic counseling, or prenatal diagnostic services.

(Added by Stats 1995, c. 415 (S.B.1360), § 4.)

Historical and Statutory Notes

Derivation: Former §10801, added by Stats.1982, c. 204, § 1, amended by Stats.1989, c. 8, § 2; Stats.1990, c. 122, § 3.

§ 103835. Scope of program; assessment of resources

The birth defects monitoring program shall operate statewide. It is the intent of the Legislature that the adequacy of program resources shall be assessed annually, and that the annual assessment shall include a consideration of at least all the following factors:

- (a) The numbers of births in the state.
- (b) The scope of program activities.
- (c) Any urgent situation requiring extraordinary commitment of present or planned program staff or resources

(Added by Stats.1995, c. 415 (S.B. 1360), § 4.)

Historical and Statutory Notes

Derivation: Former § 10802, added by Stats.1982, c. 204, § 1, amended by Stats.1990, c. 122, § 4.

§ 103840. Investigative studies

The director shall use the information collected pursuant to Section 103830 and information available from other reporting systems and health providers to conduct studies to investigate the causes of birth defects, stillbirths, and miscarriages and to determine and evaluate measures designed to prevent their occurrence. The department's investigation of poor reproductive outcomes shall not be limited to geographic, temporal, or occupational associations, but may include investigation of past exposures.

(Added by Stats.1995, c. 415 (S.B.1360), § 4.)

Historical and Statutory Notes

Derivation: Former § 10803, added by Stats.1982, c. 204, § 1, amended by Stats.1990, c. 122, § 7.

§ 103845. Advisory committee; membership

The director shall appoint an advisory committee to advise on the implementation of this chapter. Each of the disciplines of epidemiology, hospital administration, biostatistics, maternal and child health and public health shall be represented on the committee. At least one of

the members shall be a representative of the manufacturing industry.

(Added by Stats.1995, c. 415, (S.B.1360), § 4.)

Historical and Statutory Notes

Derivation: Former § 10804, added by Stats.1982, c. 204, § 1, amended by Stats.1990, c. 122, § 8.

§ 103850. Confidentiality of information; research; review and approval; civil penalty

- (a) All information collected and analyzed pursuant to this chapter shall be confidential insofar as the identity of the individual patient is concerned and shall be used solely for the purposes provided in this chapter. Access to the information shall be limited to authorized program staff, and persons with a valid scientific interest, who meet qualifications as determined by the director, who are engaged in demographic, epidemiological or other similar studies related to health, and who agree, in writing, to maintain confidentiality.
- (b) The department shall maintain an accurate record of all persons who are given access to the information in the system. The record shall include: the name of the person authorizing access; name, title, and organizational affiliation of persons given access; dates of access; and the specific purpose for which information is to be used. The record of access shall be open to public inspection during normal operating hours of the state department.
- (c) All research proposed to be conducted by persons other than program staff, using the information in the system, shall first be reviewed and approved by the director and the State Committee for the Protection of Human Subjects. Satisfaction of the terms of the director's rules for data access shall be deemed to establish a valid scientific interest for purposes of subdivision (a), entitling the researcher to review records collected pursuant to Section 103830 and to contact case subjects and controls.
- (d) Whenever program staff, pursuing program objectives, deems it necessary to contact case subjects and controls, program staff shall submit a protocol describing the research to the director and to the State Committee for the Protection of Human Subjects. Once

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a protocol is approved by that committee, program staff shall be deemed to have established a bona fide research purpose, and shall be entitled to complete the approved project and contact case subjects and controls without securing any additional approvals or waivers from any entity.

- (e) Nothing in this section shall prohibit the publishing by the department of statistical compilations relating to birth defects, stillbirth, or miscarriage that do not in any way identify individual cases or individual sources of information.
- (f) Any person who, in violation of a written agreement to maintain confidentiality, discloses any information provided pursuant to this section, or who uses information provided pursuant to this section in a manner other than as approved pursuant to this section may be denied further access to any confidential information maintained by the department. That person shall also be subject to a civil penalty of five hundred dollars (\$500). The penalty provided in this section shall not be construed as restricting any remedy, provisional or otherwise, provided by law for the benefit of the department or any person.

(Added by Stats.1995, c. 415, (S.B.1360), § 4.)

Historical and Statutory Notes

Derivation: Former § 10805, added by Stats.1982, c. 204, § 1, amended by Stats.1989, c. 8, § 4; Stats.1990, c. 122, § 9.

Library References

Records key 31. WESTLAW Topic No. 326. C.J.S. Records § § 74 to 92.

§ 103855. Contract for establishment and implementation of program

The department may enter into a contract for the establishment and implementation of the birth defects monitoring program. The contract shall include provisions requiring full compliance with all the requirements of this chapter. The term of the contract may be in excess of one year, but no longer than three years. Funds shall be allocated in accordance with the state Budget Act. Funds withheld from the contractor at the conclusion of a fiscal year until specified tasks are completed shall be released promptly on proof of

substantial completion, and shall not be offset against any funding for the subsequent fiscal year.

(Added by Stats.1995, c. 415, (S.B.1360), § 4.)

Historical and Statutory Notes

Derivation: Former § 10806, added by Stats.1985, c. 1147, § 2, amended by Stats.1989, c. 8, § 5; Stats.1990, c. 122, § 10.

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The California Birth Defects Monitoring Program—
a public health program devoted to finding causes of birth defects—
is funded through the California Department of Health Services
and jointly operated with the March of Dimes Birth Defects Foundation.

For more information about the Program, please call (559)224-2212.

Appendix 2.1.3

New Jersey Legislation

REGISTRATION - VITAL STATISTICS 26:8-40.22

26:8-40.21. Birth defects registry

The State Department of Health shall establish and maintain a birth defects registry which shall contain a confidential record of all birth defects that occur in New Jersey and any other information that the department deems necessary and appropriate in order to conduct thorough and complete epidemiologic surveys of birth defects that occur in this State and plan for and provide services to children with birth defects and their families.

L1983, c.291, § 2.

Historical Note

Effective date, see Historical Note under § 26:8-40.20.

Library References

Health and Environmental 34. C.J.S. Health and Environmental § 41.

26:8-40.2. Confidential reports of abortions of fetus with or infant affected by birth defects

- a. The Commissioner of Health, in consultation with the Public Health Council, shall require the confidential reporting to the Department of Health of all cases where a pregnancy results in a naturally aborted fetus or infant affected by a birth defect, and an electively aborted fetus that exhibits or is known to have a birth defect after 15 weeks of gestation. The reporting requirement shall apply to all infants from birth through one year of age.
- b. The Commissioner of Health shall determine the health care providers and facilities which shall be required to report all birth defects, the types of conditions or defects that shall be reported, the type of information that shall be contained in the confidential reprot and the method for making the report. In reports concerning all fetuses with anomalies, the name of the mother shall not be submitted.

Historical Note

Efective date, see Historical Note under § 26:8-40.20.

Library References

Health and Environmental 34. C.J.S. Health and Environmental § 41.

26:8-40.23 Confidentiality of reports

The confidential reports made pursuant to this act are to be used only by the Department of Health and other agencies that may be designated by the Commissioner of Health and shall not otherwise be divulged or made public so as to disclose the identity of any person to whom they relate; and to that end, such reports shall not be included under materials available to public inspection pursuant to P.L. 1963, c.73 (C.47:1A-1 et seq.).

L.1983, c.291, § 4.

Historical Note

Effective date, see Historical Note under § 26:8-40.20.

Library References

Health and Environment 34.
Records 30 et seq., 50 et seq.
C.J.S. Health and Environment § 41.
C.J.S. Records §§ 34 to 38.

26:8-40.24. Nonliability for divulging confidential information

No individual or organization providing information to the Department of Health in accordance with this act shall be deemed to be or held liable for divulging confidential information.

L.1983, c.291, § 5.

Historical Note

Effective date, see Historical Note under § 26:8-40.20.

Library References

Health and Environmental 34. Records 30 et seq., 50 et seq. C.J.S. Health and Environmental § 41. C.J.S. §§ 34 to 38.

26:8-40.25. Act not to be construed to compel submission to medical examination or to supervision by department of health

Nothing in this act shall be construed to compel any individual to submit to a medical examination or to Department of Health supervision.

L.1983, c.291, §§ 6.

Historical Note

Effective date, see Historical Note under § 26:8-40.20.

Library References

Health and Environmental 34. C.J.S. Health and Environmental § 41.

26:8-40.26. Rules and regulations

The Commissioner of Health shall promulgate rules and regulations necessary to effectuate the purposes of this act.

L.1983, c.291, § 7.

Historical Note

Effective date, see Historical Note under § 26:8-40.20.

Library References

Administrative Law and Procedure 381 et. Seq. Health and Environment 7(3), 20, 39. C.J.S. Health and Environmental §§ 2 to 13, 40 to 51, 62 to 64, 106, 125 to 137, 155, 156. C.J.S. Public Administrative Law and Procedure §§ 87 to 91.

CHAPTER 20

BIRTH DEFECTS REGISTRY

N.J.S.A. 26:8-40 et seq., specifically 26:8-40.26.

Source and Effective Date

R.2000 d.99, effective February 10, 2000. See: 31 N.J.R. 2863(a), 32 N.J.R. 802(a).

Executive Order No. 66(1978) Expiration Date

Chapter 20, Birth Defects Registry, expires on February 10, 2005.

Chapter Historical Note

Chapter 20, Birth Defects Registry, was adopted as R.1985 d.92, effective March 4, 1985. See: 16 N.J.R. 3118(a), 17 N.J.R. 591(a).

Pursuant to Executive Order No. 66(1978), Chapter 20, Birth Defects Registry, was readopted as R.1990 d.187, effective March 2, 1990. See: 21 N.J.R. 3636(a), 22 N.J.R. 1134(c).

Pursuant to Executive Order No. 66(1978), Chapter 20, Birth Defects Registry, was readopted as R.1995 d.182, effective March 2, 1995. See: 27 N.J.R. 269(a), 27 N.J.R. 1410(b).

Pursuant to Executive Order No. 66(1978), Chapter 20, Birth Defects Registry, was readopted as R.2000 d.99, effective February 10, 2000. See: Source and Effective Date. See, also, section annotations.

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SUBCHAPTER 1. LIVE BIRTHS

8:20-1.1 Definitions 8:20-1.2 Reporting requirements

SUBCHAPTER 1. LIVE BIRTHS

8:20-1.1 Definitions

The following words and terms when used in this document shall have the following meanings unless the context clearly indicates otherwise.

"Birth defect" means an abnormality of the body's structure or inherent function which is present at birth, whether such abnormality is manifest at the time of delivery or becomes apparent later in life.

"Infant" means a child from birth to one year of age.

8:20-1.2 Reporting requirements

(a) Any infant who is born to a resident of the State of New Jersey, or who becomes a resident of the State before one year of age, and who is diagnosed as having a birth defect either at birth or any time during the first year of life shall be reported to the State Department of Health and

Senior Services, Special Child, Adult and Early Intervention Services Program as follows:

- 1. The conditions listed as Congenital Anomalies (Diagnostic Codes 740.00 through 759.90) in the most recent revision of the International Classification of Diseases, Clinical Modification, shall, except as specified in (a)1ii below, be reported to Special Child, Adult and Early Intervention Services. In addition, there are several other conditions considered to be defects that are not listed under Diagnostic Codes 740.00 through 759.90 which describe Congenital Anomalies. The birth defects listed in (a)1i below shall also, in every case, be reported to Special Child, Adult and Early Intervention Services. The minor conditions listed in (a)1ii below shall not be reported to Special Child, Adult and Early Intervention Services in every case, but only as required in (a)1iii, iv and v
 - i. Congenital anomalies, including, but not limited to, the following:
 - (1) Anencephalus and similar anomalies, such as craniorachischis and inencephaly.
 - (2) Spina bifida with and without mention of hydrocephalus.
 - (3) Other congenital anomalies of the nervous system, such as: encephalocele; microcephalus; reduction deformities of the brain; congenital hydrocephalus; congenital cerebral palsies, congenital muscular dystrophies; and other anomalies, congenital diseases, lesions and any other deformities of the brain, nervous system or spinal cord.
 - (4) Congenital anomalies of the eye, such as: anophthalmos; microphthalmos; buphthalmos; congenital cataract and lens anomalies; coloboma and other anomalies of the anterior or posterior segment; congenital anomalies of eyelids, lacrimal system and orbit; and any other anomalies of the eye.
 - (5) Congenital anomalies of the ear, face and neck, such as: anomalies of the ear causing impairment of hearing; accessory auricle and any other anomalies of the ear; branchial cleft cyst or fistula; preauricular sinus; webbing of the neck; and any other anomalies of face and neck.
 - (6) Bulbus cordis anomalies and anomalies of cardiac septal closure such as: common truncus; transposition of great vessels; Tetralogy of Fallot; common ventricle; ventricular septal defect; ostium secundum type atrial septal defect; endocardial cushion defects; cor biloculare; and any other defects of septal closure.
 - (7) Other congenital anomalies of the heart, such as: anomalies of pulmonary valve; congenital tricus-

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pid atresia and stenosis; Ebstein's anomaly; congenital stenosis of aortic valve; congenital mitral stenosis of aortic valve; congenital mitral stenosis or insufficiency; hypoplastic left heart syndrome; and any other structural anomalies of the heart.

- (8) Other congenital anomalies of circulatory system, such as: patent ductus arteriosus (only in infants larger than 2,500 grams); coarctation of aorta and other anomalies of the aorta, aortic arch or atresia and stenosis of the aorta; anomalies of pulmonary artery; anomalies of great veins, absence or hypoplasia of umbilical artery; other anomalies of peripheral vascular system; or other unspecified anomalies of circulatory system.
- (9) Congenital anomalies of respiratory system, such as: choanal atresia; other anomalies of nose; webbing of larynx; other anomalies of larynx, trachea and bronchus; congenital cystic lung; agenesis, hypoplasia and dysplasia of lung; other anomalies of the lung; and other unspecified anomalies of respiratory system.
 - (10) Cleft palate and cleft lip.
- (11) Other congenital anomalies of upper alimentary tract, such as: anomalies of the tongue; anomalies of mouth and pharynx; tracheoesophageal fistula, esophageal atresia, and stenosis and other anomalies of esophagus; congenital hypertrophic pyloric stenosis, congenital hiatal hernia; other anomalies of stomach; and other unspecified anomalies of upper alimentary tract.
- (12) Other congenital anomalies of digestive system, such as: Meckel's diverticulum; atresia and stenosis of small intestine, large intestine, rectum and anal canal; Hirschsprung's disease and other congenital functional disorders of colon; anomalies of intestinal fixation; other anomalies of intestine, gall bladder, bile ducts, liver and pancreas; disorders of tooth formation, development and eruption, dentofacial anomalies, and other unspecified anomalies of the digestive system.
- (13) Congenital anomalies of genital organs, such as: anomalies of ovaries, fallopian tubes and broad ligaments; doubling of uterus and other anomalies of uterus; anomalies of cervix, vagina and external female genitalia; undescended testicle; hypospadias and congenital chordee; indeterminate sex and pseudohermaphroditism; and other unspecified anomalies of the genital system.
- (14) Congenital anomalies of urinary system, such as: renal agenesis and dysgenesis; cystic kidney disease; obstructive defects of renal pelvis and ureter; other anomalies of kidney and ureter; exstrophy of urinary bladder; atresia and stenosis of urethra and bladder neck; anomalies of urachus; other anomalies of bladder and urethra; and other unspecified anomalies of the urinary system.

- (15) Certain congenital musculoskeletal deformities, such as: of skull, face and jaw; of sternocleidomastoid muscle; of spine; congenital dislocation of hip; congenital genu recurvatum and bowing of long bones of leg; varus and valgus deformities of feet; other congenital deformities of feet such as talipes cavus, calcaneus or equinus; and other specified nonteratogenic anomalies such as pectus excavatum, pectus carinatum; club hand; congenital deformity of chest wall; dislocation of elbow; generalized flexion contractures of lower limbs; spade-like hand.
- (16) Other congenital anomalies of limbs, such as: syndactyly; reduction deformities of upper limb; reduction deformities of lower limb; other anomalies of upper limb, including shoulder girdle; and other anomalies of lower limb, including pelvic girdle.
- (17) Other congenital musculoskeletal anomalies, such as: anomalies of skull and facial bones; anomalies of spine; cervical rib; other anomalies of ribs and sternum; chondrodystrophy; osteodystrophies; anomalies of diaphragm; anomalies of abdominal wall such as prune belly syndrome; other specified anomalies of muscle, tendon, fascia and connective tissue; and other unspecified anomalies of musculoskeletal system.
- (18) Congenital anomalies of the integument, significant anomalies of skin, subcutaneous tissue, hair, nails and breast, such as birthmarks or nevi measuring four inches or greater in size, multiple skin tags (more than five in number).
- (19) Chromosomal anomalies, such as: Down's syndrome; Patau's syndrome; Edwards' syndrome; autosomal deletion syndromes and other conditions due to autosomal anomalies; gonadal dysgenesis; Klinefelter's syndrome; and other conditions due to sex chromosome anomalies or anomalies of unspecified chromosome.
- (20) Other and unspecified congenital anomalies, such as: anomalies of spleen, situs inversus; conjoined twins; tuberous sclerosis; other hamartoes; multiple congenital anomalies; and other congenital anomalies including congenital malformation syndromes affecting multiple organ systems including Laurence-Moon-Biedl syndrome, Marfan's syndrome and Prader-Willi syndrome.
- (21) Certain endocrine, nutritional and metabolic diseases and immunity disorders, includes congenital hypothyroidism; congenital hypoparathyroidism; hypopituitarism; diencephalic syndrome; adrenogenital syndrome; testicular feminization syndrome; phenylketonuria; albinism; maple syrup urine disease; argininosuccinic aciduria; glycogen storage diseases; cystic fibrosis; alpha–1 antitrypsin deficiency; DiGeorge's syndrome; congenital deficiencies of humoral immunity; cell-mediated immunity; combined immunity deficiencies; and other specified and unspecified disorders of the immune mechanisms.

- (22) Certain diseases of the blood and blood forming organs, includes hemolytic diseases of the newborn; G-6PD deficiency; hemophilia (all types); Von Willebrand's disease; and sickle-cell anemia or other hemoglobin-apathies.
- (23) Certain diseases of the nervous system and sense organs, includes hereditary and degenerative diseases of the central nervous system such as Tay Sachs disease and familial degenerative CNS diseases; Werdnig-Hoffmann disease; cerebral palsy; Moebius syndrome; hereditary retinal dystrophies, and chorioretinitis.
- (24) Certain diseases of the circulatory system, includes endocardial fibroelastosis; congenital Wolfe-Parkinson-White syndrome; and Budd-Chiari syndrome.
- (25) Certain diseases of the digestive system, includes abnormalities of jaw size, micrognathiae and macrognathia; congenital inguinal hernia with gangrene (only in females), congenital, inguinal hernia with obstruction with no mention of gangrene (only in females), congenital, inguinal hernia without obstruction with no mention of gangrene (only in females), umbilical hernia (only if not covered by skin), epigastric hernia.
- (26) Certain complications of pregnancy childbirth, and the puerperium, includes amniotic bands, amniotic cyst.
- (27) Certain diseases of the skin and subcutaneous tissue, pilonidal sinus.
- (28) Certain conditions originating in the perinatal period, includes fetal alcohol syndrome, probable fetal alcohol syndrome (includes facies), fetal alcohol effects, fetal hydantoin (Dilantin) syndrome, bronchopulmonary dysplasia, unspecified TORCH infection and certain congenital infections including congenital syphilis, congenital rubella, cytomegalovirus, toxoplasmosis, hepatitis, herpes simplex.
- (29) Neoplasms, includes lipomas of skin and subcutaneous tissue of face and other skin and subcutaneous tissue, intrathoracic and intra-abdominal organs, spermatic cord, other specified sites, lumbar, sacral, paraspinal, and other unspecified sites; benign neoplasms of skin includes blue nevus, pigmented nevus (include if greater than four inches in diameter), papilloma, dermatofibroma, syringoadenoma, dermoid cyst, hydrocystoma, syringoma; other benign neoplasms of lip, eyelid, ear, external auditory canal, skin and other unspecified parts of face, scalp, skin of neck, skin of trunk, skin of upper limb, skin of lower limb, other specified and unspecified sites including hairy nevus; hemangioma (include if: greater than four inches in diameter, multiple, more than five in number or cavernous hemangioma) of skin and subcutaneous tissue, intracranial, intra-abdominal cystic

hygroma; lymphangioma of any site, hemangioma of other and unspecified site; and certain malignant neoplasms including Wilm's tumor, retinoblastoma, other congenital neoplasms including neuroblastoma, medulloblastoma, teratoma, fibrosacrcoma, histiocytosis (malignant), neurofibromatosis.

ii. Minor conditions, as follows:

Accessory auricle Accessory nipple (supernumerary nipple, or skin tag) Anal fissure-never a defect Anal tags Bat ear Bell's Palsy Bent nose, deviation of septum Big lips Blue sclera (babies <2500 grams) Brachial palsy Breast Hypertrophy—never a defect Cafe-au-lait spots (register if five or more) Caput succedaneum Cardiac murmur 1 Cauliflower ear CNS hemorrhage Cephalhematoma-never a defect Cervical rib Chalasia (gastroesophageal reflux)—never a defect Clinodactyly (incurving of fifth finger) Congenital hydrocele Conjunctivitis-never a defect Cryptorchidism (undescended testicle) 2 Darwin's tubercle Diastasis recti-never a defect Downward eyeslant (antimongoloid) Ear tags, preauricular tags Elfin ear Epicanthal folds Epulis-never a defect Erb's palsy Erythema toxicum Esotropia Exotropia Facial palsy Flammeus nevus or port wine stain (<four inches in diameter) Flat bridge or nose Fontanel (large or small) Fractured clavicle Fused eyelids (not a defect if birth weight is <1001 Gastroesophageal reflux-never a defect Gum cysts-includes epulis, ranula, mucocele-never a defect Hemangioma—<four inches in diameter 3 Hepatomegaly Hipclick-without follow-up or therapy-not a defect Hydrocele Hydrocephaly; acquired Hymenal tags Hypoglycemia, idiopathic Hypoplastic scrotum Imperforate hymen Incurving finger (clinodactyly) Inguinal hernia in male (Note: do not report in females)

Infant of a diabetic mother; asymptomatic

Intussusception

Lanugo, excessive or persistent

Large fontanel

Laryngomalacia or tracheomalacia-never a defect

Long fingers and/or toes

Lop ear

Low set ears

Macrocheilia (big lips) Meckel's diverticulum

Meconium peritonitis

Meconium plug

Meconium stained skin or nails-never a defect

Metatarus adductus-never a defect

Metatarus varus

Microcheilia (small lips)

Mongolian spots

Mucocele-never a defect

Nasal lacrimal duct obstruction

Nail defects

Natal teeth

Neonatal acne-never a defect

Nystagmus

Orthopedic positional anomalies 4

Overlapping toes

Overriding (overlapping) sutures—never a defect

Partial syndactyly second and third toes-web extends

<one-third length of second toe

Patent ductus arteriosus (PDA) in infants <2500

grams or resolved prior to or at discharge

Patulous lips (wide lips)

Persistent fetal circulation

Petechiae—never a defect Phimosis—never a defect

Pilonidal dimple

Pilonidal cyst

Pixie-like ear

Pneumothorax

Pointed ear

Polydactyly (postaxial, type B)-skin tags on hands or

feet

Posteriorly rotated ears

Preauricular sinus

Pylorospasm (intermittent)

Ranula—never a defect Rectal fissure

Redundant foreskin Rockerbottom feet

Sacral dimple

Sebaceous cysts

Simian crease (transverse palmar crease)

Single umbilical artery

Skin cysts

Small fontanel

Small lips

Splenomegaly

Thymic hypertrophy

Tibial torsion

Tongue-tie

Torsion of spermatic cord

Torsion of testes

Tracheomalacia-never a defect

Umbilical cord atrophy

Umbilical hernias (completely covered by skin) Undescended testicle ²

Upturned nose

Upward eyeslant (mongoloid)

Vaginal cysts Vaginal tags Webbing of neck Wide nasal bridge Widely spaced nipples Widely spaced first and second toes

- iii. If a condition or defect listed in (a)1ii above appears as a single defect, a registration form shall not be completed.
- iv. If two or more of the conditions listed in (a)1ii above appear, a registration form shall be completed.
- v. If a condition or defect listed in (a)1ii above accompanies a condition or defect listed in either Diagnostic Codes 740.00 through 759.90 in the most recent revision of the International Classification of Diseases, Clinical Modification, or in (a)1i above, a registration form shall be completed.
- (b) Any live born infant with a birth defect who has not been previously registered and has expired shall be reported. Such reports shall indicate that the infant has expired.
- (c) The administrative officer of every health care facility shall be responsible for establishing the reporting procedures for that facility. The reporting procedures must insure that every infant who is initially diagnosed as having a birth defect shall be reported to the Department. All presumptive, tentative, pending, or rule out diagnoses will be reported at the time of discharge, if the child will be diagnosed at a later time or if test results are pending.
- (d) Every physician, dentist, certified nurse midwife, advanced practice nurse, and other health care professionals who diagnose or confirm birth defects shall report to the Department each infant diagnosed as having a birth defect not known to be previously reported.
- (e) The director of every clinical laboratory shall report to the Department results of postmortem examination from any infant indicating the existence of a birth defect, not known to be previously reported.
- (f) The information to be reported shall be provided upon forms supplied by the State Department of Health and Senior Services:

Special Child, Adult and Early Intervention Services

PO Box 364

Trenton, New Jersey 08625-0364

(g) The reports made pursuant to these rules are to be used only by the Department of Health and Senior Services and other agencies that may be designated by the Commissioner of Health and Senior Services and shall not otherwise be divulged or made public so as to disclose the identity of any person; and such reports shall be included under materials available to public inspection pursuant to P.L. 1963, c.73 (N.J.S.A. 47:1A-1 et seq.).

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- (h) Cytogenetic laboratories shall report the results of all postnatal chromosomal abnormalities.
- (i) When a live infant is registered, the Department shall inform the parent or legal guardian of the registration.
- (j) Every health care facility and independent clinical laboratory shall allow access to, or provide necessary information on infants with birth defects and other patients specified by characteristics for research studies related to birth defects conducted by the State Department of Health and Senior Services and which have been approved by the State Commissioner of Health and Senior Services after appropriate review for assuring protection of human subjects by the Department's Institutional Review Board. This shall include patients who came under the care of the health facility prior to March 4, 1985.
- (k) Any agency designated by the Commissioner to receive reports pursuant to this chapter shall provide to Special Child, Adult and Early Intervention Services any updated diagnostic and/or demographic information.

Amended by R.1987 d.361, effective September 8, 1987. See: 19 N.J.R. 909(b), 19 N.J.R. 1642(b). Subsection (a) added a list of congenital anomalies and other conditions which also constituted reportable birth defects.

Amended by R.1990 d.187, effective April 2, 1990.

See: 21 N.J.R. 3636(a), 22 N.J.R. 1134(c).

Reporting requirements for certain conditions specified further; reporting requirements for sickle-cell anemia and other hemoglobinopathies added; all presumptive, tentative, pending and rule out diagnoses to be reported at discharge; cytogenic laboratories to report postnatal chromosomal abnormality test results to the Department.

Amended by R.1991 d.414, effective August 5, 1991.

See: 23 N.J.R. 820(a), 23 N.J.R. 2335(a). In (a)1, added ii. through v. Amended by R.1992 d.184, effective April 20, 1992. See: 24 N.J.R. 171(a), 24 N.J.R. 1494(b).

Minor conditions added at (a)1ii.

Amended by R.2000 d.99, effective March 6, 2000.

See: 31 N.J.R. 2863(a), 32 N.J.R. 802(a).

In (a) and (f), substituted references to Special Child, Adult and Early Intervention Services for references to Special Child Health Services; rewrote (d); in (j), inserted a reference to the Department's Institutional Review Board at the end of the first sentence, and substituted a reference to March 4, 1985 for a reference to the effective date of the regulations at the end of the last sentence; and added (k).

- ¹ Do not register innocent or functional murmurs: register only if there is a definitive cardiac anomaly or register as a rule out condition if the cause of murmur is not identified at the time of discharge.
 - ² Register only if there is clinical evidence of congenital absence.
- ³ Register cavernous hemangiomas and multiples of five or more.
- ⁴ Do not register if defect can be corrected passively and does not require casting or bracing.

Appendix 2.1.4

New York Legislation

Pertinent Public Health Laws and Regulations

The following laws and regulation establish the legal authority to collect information on birth defects and genetic diseases, to perform studies, and to maintain the confidentiality of the information and limits its use to research and the improvement of quality of care.

Section 206 (1) of the Public Health Law

- 1. The Commissioner shall:
 - (d) investigate the causes of disease, epidemics, the sources of mortality, and the effects of localities, employments and other conditions, upon the public health;
 - (e) obtain, collect and preserve such information relating to marriage, birth, mortality, disease and health as may be useful in the discharge of his duties or may contribute to the promotion of health or the security of life in the state,
 - (i) cause to be made such scientific studies and research which have for their purpose the reduction of morbidity and mortality and the improvement of the quality of medical care through the conduction of medical audits within the state. In conducting such studies and research, the commissioner is authorized to receive reports on forms prepared by him and the furnishing of such information to the commissioner, or his authorized representatives, shall not subject any person, hospital, sanitarium, rest home, nursing home, or other person or agency furnishing such information to any action for damages or other relief. Such information when received by the commissioner, or his authorized representatives, shall be kept confidential and shall be used solely for the purposes of medical or scientific research or the improvement of the quality of medical care through the conduction of medical audits. Such information shall not be admissible as evidence in any action of any kind in any court or before any other tribunal, board, agency or person.

Section 225(5)(t) of the Public Health Law.

- 5. The Sanitary code may:
 - (t) facilitate epidemiological research into the prevention of environmental diseases, when such research is conducted pursuant

to paragraph (j) of subdivision one of section two hundred six of this chapter, by establishing regulations designating as environmentally related diseases those pathological conditions of the body or mind resulting from contact with toxins, or teratogens in solid, liquid or gaseous form, or in the form of ionizing radiation or nonionizing electromagnetic radiation, and by requiring the reporting of such diseases or suspected cases in such diseases to the department by physicians, medical facilities and clinical laboratories. Any information provided to the department pursuant to such regulations shall be in the form required by the department and shall be kept confidential and used by the commissioner pursuant to the provisions of paragraph (j) of subdivision one of section two hundred six of this chapter, and other applicable laws relating to the confidential treatment of patient and medical data.

Section 2733 of the Public Health Law

- 1. Birth defects and genetic and allied diseases shall be reported by physicians, hospitals, and persons in attendance at birth in the manner on and such forms as may be prescribed by the commissioner.
- 2. Such reports and information shall be kept confidential and shall not be admissible as evidence in an action or proceeding in any court or before any other tribunal, board, agency or person. The commissioner may, however, publish analyses of such information from time to time for scientific and public health purposes, in such manner as to assure that the identities of the individuals concerned cannot be ascertained.

State Sanitary Code: Part 22 – Environmental Diseases (Statutory authority: Public Health Law, §§ 225 [5][t], 206 [1][j])

- 22.1 Supplementary reports of spontaneous abortions and fetal deaths for epidemiologic surveillance; filing. Every physician and hospital shall file a supplemental report with the State Commissioner of Health of each spontaneous abortion or other fetal death occurring naturally. Such report shall be filed within 10 days of the occurrence of such event on such fo9rms as may be prescribed by the commissioner to facilitate epidemiologic investigation and surveillance.
- 22.2 Supplementary reports of low birth weights for epidemiologic surveillance; filling. Every physician, hospital, and person in attendance at live births shall file a supplementary report with the State commission of Health of each live birth for which the birth weight is 2,500 grams(5.2 pounds) less.

Such report shall be filed within 10 days of the birth and shall be on such formed as may be prescribed by the commissioner to facilitate epidemiologic investigation and surveillance.

Regulation specifically establishing the CMR

- 22.3 Supplementary reports of certain congenital anomalies for epidemiological surveillance; filing. Every physician and hospital in attendance on an individual diagnosed within two years of birth as having one or more of the congenital anomalies listed in this section shall file a supplementary report with the State Commissioner of Health within 10 days of diagnosis thereof. Such report shall be on such forms as may be prescribed by the commissioner to facilitate epidemiological investigation and surveillance.
- 22.9 Reports: place of filing. All reports required by this part shall be filed with the Director of the Bureau of Environmental Epidemiology and Occupational Health, Division of Epidemiology, New York State Department of Health, Empire State Plaza, Tower Building, Albany, NY 12237.

Appendix 2.1.5 Oklahoma Legislation

PUBLIC HEALTH AND SAFETY

63 § 1-550.2

Historical and Statutory Notes

The repealed section, derived from Laws 1985, c 60, § 1; Laws 1986, c. 49, § 1, related to the reporting of cases of birth defects.

See, now, §§ 1-550.1, 1-550.2 of this title.

§ 1-550.1 Definitions

As used in this act:1

- 1. "Birth defect" means any physical or chemical abnormality present at birth;
- 2. "Commissioner" means the Commissioner of Health;
- "Department" means the Oklahoma State Department of Health;
- 4. "ICD-9-CM diagnostic code categories" means the International Classification
- of Disease which assigns numbers to each of the congenital anomalies; and
- "Poor reproductive outcomes" includes but is not limited to stillbirths and miscarriages.

Added by Laws 1987, c. 199, § 1, eff. Nov. 1, 1987.

1 Section 1-550.1 et seq. of this title.

Historical and Statutory Notes

Section 4 of Laws 1987, c. 199 provides for an effective date.

§ 1-550.2 Birth defects surveillance program

- A. It is hereby found that the occurrence of a birth defect is a tragedy for the child, the family and the community, and a matter of vital concern to the public health. A system to obtain more information about these conditions could result in their prevention, treatment and management. Therefore, it is the intent of the Oklahoma State Legislature, in enacting this section, to:
 - 1. Obtain information on the incidence and trends of birth defects and poor reproductive outcomes;
- Obtain information to determine whether environmental hazards are associated with birth defects and poor reproductive outcomes;
- 3. Obtain information as to other possible causes of birth defects and poor reproductive outcomes;
- 4. Develop prevention strategies for reducing the incidence of birth defects, and poor reproductive outcomes.
- B. The Commissioner of Health may establish a system for the collection and verification of information concerning birth defects and other poor reproductive outcomes. In establishing the system, the Commissioner may require general acute care hospitals to maintain a list of patients up to six (6) years of age who have been diagnosed with birth defects incorporated within the ICD-9-CM diagnostic code categories 740 through 759.9 or such other information as the Commissioner deems appropriate, and all women discharged with a diagnosis of stillbirth or miscarriage. The list shall be made available to the Commissioner upon request and shall be used solely for purposes provided in this section.
- C. The Commissioner may require general acute care hospitals, and other sources as deemed necessary, to make available to the State Department of Health the medical records of those patients who have been diagnosed with birth defects or poor reproductive outcomes as required in this section.
 - D. The system shall be implemented statewide.
- E. The Commissioner may use the information collected pursuant to subsection B of this section and information available from other reporting systems and health providers to conduct studies to:
 - 1. Investigate the causes of birth defects and poor reproductive outcomes;

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PUBLIC HEALTH AND SAFETY

- 2. Determine and evaluate measures to prevent their occurrences; and
- 3. Where possible ensure delivery of services for children identified with birth defects. The Department's investigation of poor reproductive outcomes shall include geographic, time-related or occupational associations, as well as investigations of past exposure to potentially harmful substances.
- F. Where Commissioner may appoint an advisory committee of health professionals who shall advise on the implementation of this section. Advisory committee members shall serve without compensation.
- G. If the Commissioner finds it is necessary to collect information from sources other than general acute care hospitals, the Commissioner shall first submit for approval to the advisory committee a proposal stating the need for such information.
- H. All information collected and analyzed pursuant to this section shall be confidential insofar as the identity of the individual patient is concerned and shall be used solely for the purpose provided in this section. Access to such information shall be limited to the State Department of Health, provided that the Commissioner may provide access to those scientists approved by the advisory committee who are engaged in demographic, epidemiological or other similar studies related to health, and who agree, in writing as nonstate employees, to be identified and coded while maintaining confidentiality as described herein.
- I. The Department shall maintain an accurate record of all persons who are given access to the information in the system. The record shall include:
 - 1. The name of the persons authorizing access;
 - 2. The name, title and organizational affiliation of persons given access;
 - 3. The dates of access;
 - 4. The specific purpose for which the information is to be used; and
 - 5. The results of the independent research.
- J. Nothing in this section shall prohibit the publishing of statistical complications relating to birth defects or poor reproductive outcomes which do not in any way identify individual cases or individual sources of information.
- K. Any person who, in violation of a written agreement to maintain confidentiality, willfully discloses any information provided pursuant to this section shall be denied further access to any confidential information maintained by the Department. That person shall also be deemed guilty of a misdemeanor, and upon conviction thereof shall be punished by a fine of Two Hundred Dollars (\$200.00) or imprisonment in the county jail for not more than thirty (30) days, of by both such fine and imprisonment.
- L. The State Board of Health is authorized to adopt, amend and repeal rules and regulations for the purpose of carrying out the provisions of this section.

Added by Laws 1987, c. 199, § 2, eff. Nov. 1, 1987. Amended by Laws 1992, c. 123, § 1, emerg. eff. April 23, 1992.

§ 1-551. Repealed by Laws 1987, c. 197, § 2, eff. Nov. 1, 1987

Historical and Statutory Notes

The repealed section, derived from Laws 1985, c. 60, § 2: Laws 1986, c. 49, § 2, related to the reporting cases of cancer.

See, now, § 1-551.1 of this title.

§ 1-551.1 Tumor registry

A. The State Commissioner of Health shall establish and maintain an up-to-date tumor registry to ensure an accurate and continuing source of data concerning such cancerous, precancerous and tumorous diseases as the State Board of Health may by rule specify. Such registry may include data necessary for epidemiological surveys and scientific research, and other data which is necessary and proper to further the recognition, prevention, control, treatment and cure of cancer, precancerous and tumorous diseases.

Texas Legislation

Texas legislation

HEALTH & SAFETY CODE

CHAPTER 87. BIRTH DEFECTS

SUBCHAPTER A. GENERAL PROVISIONS

Sec. 87.001. DEFINITIONS. In this chapter:

- (1) "Birth defect" means a physical or mental functional deficit or impairment in a human embryo, fetus, or newborn resulting from one or more genetic or environmental causes.
- $\mbox{(2) "Communicable disease" has the meaning assigned by Section <math>81.003.$
- (3) Repealed by Acts 1995, 74th Leg., ch. 76, Sec. 8.134, eff. Sept. 1, 1995.
- (4) "Environmental causes" means the sum total of all the conditions and elements that make up the surroundings and influence the development of an individual.
- $$\left(5\right)$$ "Harmful physical agent" has the meaning assigned by Section 503.001.
 - (6) "Health professional" means an individual whose:
- (A) vocation or profession is directly or indirectly related to the maintenance of health in another individual; and
- (B) duties require a specified amount of formal education and may require a special examination, certificate, or license or membership in a regional or national association.
 - (7) "Health facility" includes:
- (A) a general or special hospital licensed by the department under Chapter 241;
- (B) a physician-owned or physician-operated clinic;
- (C) a publicly or privately funded medical school;
- (D) a state hospital or state school maintained and managed by the Texas Department of Mental Health and Mental Retardation;
 - (E) a genetic evaluation and counseling center;
- (F) a public health clinic conducted by a local health unit, health department, or public health district organized and recognized under Chapter 121;

- (G) a physician peer review organization; and
- (H) another facility specified by board rule.
- (8) "Midwife" has the meaning assigned by Section 203.002, Occupations Code.
- (9) "Local health unit" has the meaning assigned by Section 121.004.
- (10) "Toxic substance" has the meaning assigned by Section 503.001.

Added by Acts 1993, 73rd Leg., ch. 602, Sec. 1, eff. Sept. 1, 1993. Amended by Acts 1995, 74th Leg., ch. 76, Sec. 8.134, eff. Sept. 1, 1995; Acts 2001, 77th Leg., ch. 1420, Sec. 14.774, eff. Sept. 1, 2001.

Sec. 87.002. CONFIDENTIALITY. (a) Except as specifically authorized by this chapter, reports, records, and information furnished to a department employee or to an authorized agent of the department that relate to cases or suspected cases of a health condition are confidential and may be used only for the purposes of this chapter.

- (b) Reports, records, and information relating to cases or suspected cases of health conditions are not public information under Chapter 552, Government Code, and may not be released or made public on subpoena or otherwise except as provided by this chapter.
- (c) The department may release medical, epidemiological, or toxicological information:
- (1) for statistical purposes, if released in a manner that prevents the identification of any person;
- (2) with the consent of each person identified in the information or, if the person is a minor, the minor's parents, managing conservator, guardian, or other person who is legally authorized to consent;
- (3) to medical personnel, appropriate state agencies, health authorities, regional directors, and public officers of counties and municipalities as necessary to comply with this chapter and board rules relating to the identification, monitoring, and referral of children with birth defects;
- (4) to appropriate federal agencies, such as the Centers for Disease Control of the United States Public Health

Service; or

- (5) to medical personnel to the extent necessary to protect the health or life of the child identified in the information.
- (d) A board member, the commissioner, another employee of the department, or an authorized agent may not be examined in a civil, criminal, special, or other proceeding as to the existence or contents of pertinent records of or reports or information about a child identified or monitored for a birth defect by the department without the consent of the child's parents, managing conservator, guardian, or other person authorized by law of this state or another state or by a court order to give consent.

Added by Acts 1993, 73rd Leg., ch. 602, Sec. 1, eff. Sept. 1, 1993. Amended by Acts 1995, 74th Leg., ch. 76, Sec. 5.95(88), 8.130, eff. Sept. 1, 1995.

Sec. 87.003. CONTRACTS. The department may enter into contracts or agreements with persons as necessary to implement this chapter. The contracts or agreements may provide for payment by the state for supplies, equipment, data, and data collection and other services.

Added by Acts 1993, 73rd Leg., ch. 602, Sec. 1, eff. Sept. 1, 1993.

Sec. 87.004. LIMITATION OF LIABILITY. A health professional, a health facility, or an administrator, officer, or employee of a health facility subject to this chapter is not civilly or criminally liable for divulging information required to be released under this chapter, except in a case of gross negligence or wilful misconduct.

Added by Acts 1993, 73rd Leg., ch. 602, Sec. 1, eff. Sept. 1, 1993.

Sec. 87.005. COOPERATION OF GOVERNMENTAL ENTITIES. Another state board, commission, agency, or governmental entity capable of assisting the department in carrying out the intent of this chapter shall cooperate with the department and furnish expertise, services, and facilities to the program.

Added by Acts 1993, 73rd Leg., ch. 602, Sec. 1, eff. Sept. 1, 1993.

SUBCHAPTER B. BIRTH DEFECTS MONITORING PROGRAM

Sec. 87.021. SURVEILLANCE PROGRAM; REGISTRY ESTABLISHED. (a) The board shall establish in the department a

program to:

- (1) identify and investigate certain birth defects in children; and
- (2) maintain a central registry of cases of birth defects.
- (b) The board may authorize the department to implement a statewide program or to limit the program to a part or all of one or more public health regions, depending on the funding available to the department. In establishing the program, the board shall consider:
- (1) the number and geographic distribution of births in the state;
- (2) the trained personnel and other departmental resources that may be assigned to the program activities; and
- (3) the occurrence or probable occurrence of an urgent situation that requires or will require an unusual commitment of the department's personnel and other resources.
- (c) The board and the department shall design the program so that the program will:
- (1) provide information to identify risk factors and causes of birth defects;
- (2) provide information on other possible causes of birth defects;
- (3) provide for the development of strategies to prevent birth defects;
- (4) provide for interview studies about the causes of birth defects;
- (5) together with other departmental programs, contribute birth defects data to a central registry;
- (6) provide for the appointment of authorized agents to collect birth defects information; and
- $\qquad \qquad \hbox{(7)} \quad \text{provide for the active collection of birth defects} \\ \text{information.}$
- (d) The board shall adopt rules to govern the operation of the program and carry out the intent of this chapter. At a minimum, the rules shall:
 - (1) use a medically recognized system to specify the

birth defects to be identified and investigated;

- (2) select a system for classifying the birth defects according to the public health significance of each defect to prioritize the use of resources;
- (3) develop a system to select and specify the cases to be investigated;
- (4) specify a system for selecting the demographic areas in which the department may undertake investigations; and
- (5) prescribe the training and experience a person must have for appointment as an authorized agent of the department.
- (e) In adopting the rules required by Subsection (d), the board shall consider at least:
- (1) the known incidence and prevalence rates of a birth defect in the state or portions of the state;
- (2) the known incidence and prevalence rates of a particular birth defect in specific population groups who live in the state or portions of the state;
- (3) the morbidity and mortality resulting from the birth defect; and
- (4) the existence, cost, and availability of a strategy to prevent and treat the birth defect.
- (f) In addition to providing for the active collection of birth defects information under Subsection (c)(7), the board and the department may design the program to also provide for the passive collection of that information.

Added by Acts 1993, 73rd Leg., ch. 602, Sec. 1, eff. Sept. 1, 1993. Amended by Acts 2003, 78th Leg., ch. 1042, Sec. 1, eff. June 20, 2003.

Sec. 87.022. DATA COLLECTION. (a) To ensure an accurate source of data necessary to investigate the incidence, prevalence, and trends of birth defects, the board may require a health facility, health professional, or midwife to make available for review by the department or by an authorized agent medical records or other information that is in the facility's, professional's, or midwife's custody or control and that relates to the occurrence of a birth defect specified by the board.

(b) The board by rule shall prescribe the manner in which

records and other information are made available to the department.

(c) The board shall adopt procedural rules to facilitate cooperation between the health care facility, health professional, or midwife and a department employee or authorized agent, including rules for notice, requests for medical records, times for record reviews, and record management during review.

Added by Acts 1993, 73rd Leg., ch. 602, Sec. 1, eff. Sept. 1, 1993.

Sec. 87.023. REFERRAL FOR SERVICES. A child who meets the medical criteria prescribed by board rule, and the child's family, shall be referred to the department's case management program for guidance in applying for financial or medical assistance available through existing state and federal programs.

Added by Acts 1993, 73rd Leg., ch. 602, Sec. 1, eff. Sept. 1, 1993.

SUBCHAPTER C. INVESTIGATIONS AND INSPECTIONS

Sec. 87.041. INVESTIGATIONS. (a) The department may conduct investigations, including epidemiological or toxicological investigations, of cases of specified birth defects.

- (b) The department may conduct these investigations to determine the nature and extent of the disease or the known or suspected cause of the birth defect and to formulate and evaluate control measures to protect the public health. The department's investigation is not limited to geographic, temporal, or occupational associations and may include investigation of past exposures.
- (c) A person shall provide medical, demographic, epidemiological, toxicological, and environmental information to the department under this chapter.
- (d) A person is not liable in damages or other relief for providing medical or other confidential information to the department during an epidemiological or toxicological investigation.

Added by Acts 1993, 73rd Leg., ch. 602, Sec. 1, eff. Sept. 1, 1993.

Sec. 87.042. DEPARTMENTAL INVESTIGATORY POWERS. To conduct an investigation under this chapter, the commissioner or the commissioner's designee has the same authority to enter, inspect, investigate, and take samples and to do so in the same manner as is provided for communicable diseases under Sections

81.061, 81.063, 81.064, and 81.065.

Added by Acts 1993, 73rd Leg., ch. 602, Sec. 1, eff. Sept. 1, 1993. Amended by Acts 1995, 74th Leg., ch. 76, Sec. 8.132, eff. Sept. 1, 1995.

SUBCHAPTER D. CENTRAL REGISTRY

Sec. 87.061. REGISTRY; CONFIDENTIALITY. (a) Information collected and analyzed by the department or an authorized agent under this chapter may be placed in a central registry to facilitate research and to maintain security. The department may also store information available from other departmental programs and information from other reporting systems and health care providers.

- (b) The department shall use the registry to:
- (1) investigate the causes of birth defects and other health conditions as authorized by Texas statutes;
- (2) design and evaluate measures to prevent the occurrence of birth defects and other health conditions; and
- (3) conduct other investigations and activities necessary for the board and department to fulfill their obligation to protect the health of the public.
- (c) The department may store in the central registry information that is obtained from the section of the birth certificate entitled "For Medical and Health Use Only." This information may be used only as provided by Section 191.002(b), relating to the form and contents of the birth certificate.

Added by Acts 1993, 73rd Leg., ch. 602, Sec. 1, eff. Sept. 1, 1993.

Sec. 87.062. ACCESS TO INFORMATION. (a) Access to the central registry information is limited to authorized department employees and other persons with a valid scientific interest who are engaged in demographic, epidemiological, or other studies related to health and who agree in writing to maintain confidentiality.

- (b) The department shall maintain a listing of each person who is given access to the information in the central registry. The listing shall include:
 - (1) the name of the person authorizing access;
- (2) the name, title, and organizational affiliation of each person given access;

- (3) the dates of access; and
- $\qquad \qquad \textbf{(4)} \quad \text{the specific purpose for which the information was} \\$ used.
- (c) The listing is public information, is open to the public under Chapter 552, Government Code, and may be inspected during the department's normal hours of operation.

Added by Acts 1993, 73rd Leg., ch. 602, Sec. 1, eff. Sept. 1, 1993. Amended by Acts 1995, 74th Leg., ch. 76, Sec. 5.95(88), eff. Sept. 1, 1995.

Sec. 87.063. RESEARCH; REVIEW AND APPROVAL. (a) The commissioner and the department's committee for the protection of human subjects shall review each research proposal that requests the use of information in the central registry. The board shall adopt rules establishing criteria to be used in deciding if the research design should be approved. A proposal that meets the approval criteria is considered to establish a valid interest as required by Section 87.062(a), and the commissioner and the committee shall authorize the researcher to review the records relevant to the research proposal and to contact cases and controls.

(b) If an investigator using central registry data under a research design approved under this section believes it is necessary to contact case subjects and controls, the investigator must submit a protocol describing the purpose and method to the commissioner and the department's committee for the protection of human subjects. If the contact protocol is approved, the investigator is considered to have established a bona fide research, development, or planning purpose and is entitled to carry out the contacts without securing additional approvals or waivers from any entity.

Added by Acts 1993, 73rd Leg., ch. 602, Sec. 1, eff. Sept. 1, 1993. Amended by Acts 1995, 74th Leg., ch. 76, Sec. 8.133, eff. Sept. 1, 1995.

Sec. 87.064. REPORT OF CENTRAL REGISTRY ACTIVITIES AND FINDINGS. (a) The department shall publish an annual report of activities using data contained in the central registry. The report shall include:

- (1) a description of research projects in progress since the last report and the sponsors and principal investigators directing each project;
- (2) results of the completed research projects either as an abstract or a complete scientific paper that has been reviewed and approved by an appropriate jury;
- (3) a summary of the statistical information compiled in the registry, including a specific discussion of any clusters, high or low incidences, or prevalences or trends encountered;
- (4) any policy, research, educational, or other recommendations the department considers appropriate; and
- (5) such other information the editors of the report find is appropriate.
- $\begin{tabular}{ll} \textbf{(b)} & \textbf{The department may publish periodic reports in addition} \\ \textbf{to the annual report.} \end{tabular}$

Added by Acts 1993, 73rd Leg., ch. 602, Sec. 1, eff. Sept. 1, 1993.

Sec. 87.065. COORDINATION WITH MEXICO. In developing the central registry and conducting research in areas of this state that border Mexico, the department shall make every effort to coordinate its efforts with similar efforts and research programs in Mexico.

Added by Acts 1993, 73rd Leg., ch. 602, Sec. 1, eff. Sept. 1, 1993.

Appendix 2.2 Table of Birth Defects Legislation

Appendix 2.2 Table of State Birth Defects Legislation

	State I	Birth D	efects Legislat	ion – April 2004
State	Name of Birth Defects Surveillance Program	Leg/ Rule	Year	Citation
Alabama	Alabama Birth Defects Surveillance and Prevention Program	Yes	2001	Code of Ala. § 22-10A-2
Alaska	Alaska Birth Defects Registry (ABDR)	Yes	1996 (enact)	7AAC 27.012
Arizona	Arizona Birth Defects Monitoring Program	Yes	1988 (enacted); 1991 (adopted); 2001 (revised)	Statue: ARS § 36-133 Rule: Title 9, Chapter 4, Articles 1 &5 A.R.S. § 36-133 (2001)
Arkansas	Arkansas Reproductive Health Monitoring System	Yes	1985 (enacted); 1999 (revised)	Bill 214 (1985) A.C.A. § 20-16-201
California	California Birth Defects Monitoring Program	Yes	1982 (enacted); 1996 (recodified)	Health and Safety Code, Division 102, Part 2, Chapter 1, Sections 103825-103855, effective 1982. Recodified 1996: § 103825, 103855, 103830, 103835, 125050, 103840, 103850, 125000
Colorado	Colorado Responds To Children With Special Needs	Yes	1985 (enacted)	Colorado Revised Statutes 25-1.5-101 - 25-1.5-105
Connecticut	Connecticut Birth Defects Registry	Yes	1991 (enacted)	Sec. 10a-132b transferred to sec 19a-56a in 1999 § 19a-56a, 19a-56b (2001) State has 2 statutes which mandate the reporting of children with birth defects. They are: Sec. 19a-53. (Formerly Sec. 19-21). Reports of physical defects of children; Sec. 19a-54. (Formerly Sec. 19-21a). Registration of physically handicapped children; Sec. 19a-56a for Birth defects surveillance program, and Sec. 19a-56b, add Sec. 19a-56c Advisory committee.
Delaware	Delaware Birth Defects Surveillance Project	Yes	1997 (enacted)	House Bill No. 197, an act to amend Title 16 of Del. Code 16 Code §203 (2000); §201, §202
District of Columbia	District of Columbia Birth Defects Surveillance and Prevention Program	No		
Florida	Florida Birth Defects Registry	Yes	1999	Sec 381.0031 (1, 2) provides for a list of reportable diseases/conditions in Florida. Congenital anomalies were added in 1999.
Georgia	1) Metropolitan Atlanta Congenital Defects Program (MACDP) 2) Georgia Birth Defects Reporting and Information System (GBDRIS)	Yes	2002 (GBDRIS system)	MACDP: Official Code of GA (OCGA) 31-12-2 GBDRIS: Birth Defects reporting activated statewide in 2002; Citation: Add GA 31-1-3.2 and DHR Rules 290-5- 302 and 290-5-24
Hawaii	Hawaii Birth Defects Program	Yes	1989 (enacted) 2002 (modified)	8/15/1988 to 6/30/2002 - Hawaii Revised Statutes (HRS), Sections 321-31 and 338-2 in conjunction per Executive Chamber ruling by Governor on 6/16/1989. HRS Sections 324-1 and 324-2 for additional legislative authority (1990 Amendments). 7/1/02 to Present - Act 252 - Relating to Birth Defects (SB 2763, SD 2, HD 2, CD 1).
Idaho		No birth defects surveillance program		

State Birth Defects Legislation – April 2004					
State	Name of Birth Defects Surveillance Program	Leg/ Rule	Year	Citation	
Illinois	Adverse Pregnancy Outcomes Reporting System	Yes	1985 (enacted)	IL Health and Hazardous Substances Registry Act (410 ILCS 525); 235 ILCS 5/6-24a (2001)	
Indiana	Indiana Birth Defects and Problems Registry	Yes	2001 (enacted)	IC 16-38-4, Rule 410 IAC 21-3	
lowa	Iowa Birth Defects Registry	Yes	1983 (enacted); 2001, 2003 (revised)	IA Code 136A.1, 136A.2, 136A.3, 136A.5, 136A.6; 135.40 Administrative Code of IA 641-1.3(139A); 641-4.1, 641-4.7(136A) (revised 2001) The administrative rules for the Birth Defects Institute (which includes the Registry and other programs such as newborn screening) are undergoing revision in procedures for newborn screening & prenatal screening, but the revision does not affect the Registry's rules.	
Kansas	Birth Defects Reporting System	Yes	1979 (enacted)	KSA 65-102	
Kentucky	Kentucky Birth Surveillance Registry	Yes	1992 (enacted); 1997	KRS 211.651-211.670 (1992); HB372 KRS §211.660, §211.665, §211.670 (2001) KY SB 219 (2002)	
Louisiana	Louisiana Birth Defects Monitoring Network	Yes	2001 (enacted)	R.S. 40:31.41-40.31.48, Act No. 194 (2002)	
Maine	Maine Birth Defects Program	Yes	1999	22MRSA c. 1687	
Maryland	Maryland Birth Defects Reporting and Information System	Yes	1982	Health-General Article, Section 18-206; Annotated Code of MD	
Massachusetts	Massachusetts Center For Birth Defects Research and Prevention, Birth Defects Monitoring Program	Yes	1963 (enacted); 2002 (revised)	Massachusetts General Laws, Chapter 111, Section 67E. In 2002 the Massachusetts Legislature amended this statute, expanding the birth defects monitoring program. The new law: 1) increases mandated reporting up to age three; 2) requires physicians to report to MDPH within 30 days of diagnosis; 3) sets out requirements for the use of this data; 4) requires MDPH to promulgate regulations governing the operation of the Birth Defects Monitoring Program.	
Michigan	Michigan Birth Defects Registry	Yes	1988	Public Health Act 236 of 1988	
Minnesota	Minnesota Birth Defects Information System	Yes	2004	MS 144.2215	
Mississippi	Mississippi Birth Defects Registry	Yes	1997	Sec. 41-21-205 of MS Code	
Missouri	Missouri Birth Defects Registry	No			
Montana	Montana Birth Outcomes Monitoring System	No			
Nebraska	Nebraska Birth Defects Registry	Yes	1972	Laws 1972, LB 1203, §1, §2, §3, §4 (alternate citation: Public Health and Welfare [Codes] §71-645, §71-646, §71-647, §71-648, §71-649)	
Nevada	Nevada Birth Defects Registry	Yes	1999	NRS 442.300-442.330; regulation - NAC 442	
New Hampshire	New Hampshire Birth Conditions Program	No			
New Jersey	Special Child Health Services Registry	Yes	1983 (enacted); 2000 (readopted)	Bill 757, NJSA 26:8, NJAC 8:20 (enacted 8-4-1983 with effective date of 3-4-1985)	

		SIRTN D		ion – April 2004		
State	Name of Birth Defects Surveillance Program	Leg/ Rule	Year	Citation		
New Mexico	New Mexico Birth Defects Prevention and Surveillance System	Yes	2000 (enacted)	In January 2000, birth defects became a reportable condition. These conditions are updated by the Office of Epidemiology. This did not involve legislation, only a change in regulations.		
New York	New York State Congenital Malformations Registry	Yes	1982	Public Health Law Art. 2, Title, II, Sect 225(5)(t) and Art. 2 Title I, sect 206(1)(j): Codes, Rules and Regulations, Chap 1, State Sanitary Code, part 22.3		
North Carolina	North Carolina Birth Defects Monitoring Program	Yes	1995; 2001 (adopted rules)	15A NCAC 26C.0101-0106 NC adopted rules in 2001 to support the statute.		
North Dakota	North Dakota Birth Defects Monitoring System	Yes		ND Centry Code 50-10		
Ohio	Ohio Connections for Children with Special Needs	Yes	2000 (enacted)	House Bill 354 The legislation authorizes the state to have a birth defects system, but until funding is identified/ secured, does not require the Ohio Dept. of Health to implement a system. The document is available at: www.legislature.state.oh.us/BillText123/123_HB_534_5 _Y.htm		
Oklahoma	Oklahoma Birth Defects Registry	Yes	1992	63 O.S. Sec 1-550.2 (1992)		
Oregon	No birth defects surveillance program					
Pennsylvania	Pennsylvania Follow-Up Outreach, Referral and Education For Families	No				
Puerto Rico	Puerto Rico Folic Acid Campaign and Birth Defects Surveillance System	No				
Rhode Island	Rhode Island Birth Defects Surveillance Program	Yes	2003	House 5390, Senate 105 (Title 23, Chapter 13.3 of the General Laws)		
South Carolina	South Carolina Birth Defects Surveillance And Prevention	No				
South Dakota			No birth defects	surveillance program		
Tennessee	Tennessee Birth Defects Surveillance Project	Yes	2000	TCA 68-5-506		
Texas	Texas Birth Defects Monitoring Division	Yes	1993 (enacted)	Health and Safety Code, Title 2, Subtitle D, Section 1, Chapter 87.		
Utah	Utah Birth Defect Network	Yes	1999	Birth Defect Rule (R398-5)		
Vermont	Vermont Birth Information Network	Yes	2003	Act 32		
Virginia	Virginia Congenital Anomalies Reporting and Education system	Yes	198 (enacted); 1986, 1988 (amended)	Bill 396, HL 32.1.69.1, Art. 8 1985 (1986)		
Washington	Washington State Birth Defects Surveillance System	Yes	2004	HB1105, notifiable conditions WAC 246-101 (2000)		

State Birth Defects Legislation – April 2004				
State	Name of Birth Defects Surveillance Program	Leg/ Rule	Year	Citation
West Virginia	West Virginia Congenital Abnormalities Registry, Education and Surveillance System	Yes	1991 (enacted); 2002 (updated)	HB1747, SS Sec: 16-5-12a (1991); 16-40-1 (2002) *old legislation is still in effect but additional legislation that now calls for advisory committee to the Birth Defects Information System has been added in a different section of code. *this occurred during the 2002 session - SB 672. *new code citation is 16-40-1 *legislative rules are in process of being completed for inclusion during the 2004 session.
Wisconsin	Wisconsin Birth Defects Prevention and Surveillance Program	Yes	2003	1999 WA 114 Statute 253.12; HFS 116effective April 1, 2003
Wyoming	No birth defects surveillance program			

Definitions Used to Determine Covered Entity Status Under the Privacy Rule

Definitions Used to Determine Covered Entity Status Under the Privacy Rule

Covered Entity

A health plan, a health care clearinghouse, or a health care provider who conducts electronic transactions. These transactions are described at 45 C.F.R.164.

Health Care

Care, services, or supplies related to the health of an individual. It includes, but is not limited to, the following:

- Preventive, diagnostic, rehabilitative, maintenance, or palliative care, and counseling, service, assessment, or procedure with respect to the physical or mental condition, or functional status, of an individual or that affects the structure or function of the body
- Sale or dispensing of a drug, device, equipment, or other item in accordance with a prescription. See 45 C.F.R.160.103

Covered Transactions

Transactions for which the Secretary of Health and Human Services has adopted standards and which can be found at 45 C.F.R. Part 162. If a health care provider uses another entity (such as a clearinghouse) to conduct covered transactions in electronic form on its behalf, the health care provider is considered to be conducting the transaction in electronic form.

Required by Law

A mandate contained in law that compels an entity to use or to disclose protected health information, and that is enforceable in a court of law. Required by law includes, but is not limited to, court orders and court-ordered warrants; subpoenas or summons issued by a court, grand jury, a governmental or tribal inspector general, or an administrative body authorized to require the production of information; a civil or an authorized investigative demand; Medicare conditions of participation with respect to health care providers participating in the program; and statutes or regulations that require the production of information, including statutes or regulations that require such information if payment is sought under a government program providing public benefits.

Research

Systematic investigation, including research development, testing, and evaluation, designed to develop or contribute to generalizable knowledge.

Office of Civil Rights (OCR) HIPAA Privacy Regulation Text

Office of Civil Rights (OCR) HIPAA Privacy Regulation Text

Below is the actual text of the HIPAA privacy regulation, then the comments from the Privacy Rule's preamble, or the modification guidance issued with the Rule.

Office for Civil Rights (OCR)/HIPAA Privacy Regulation Text

§ 164.512 Uses and disclosures for which an authorization or opportunity to agree or object is not required.

A covered entity may use or disclose protected health information without the written authorization of the individual, as described in § 164.508, or the opportunity for the individual to agree or object as described in § 164.510, in the situations covered by this section, subject to the applicable requirements of this section. When the covered entity is required by this section to inform the individual of, or when the individual may agree to, a use or disclosure permitted by this section, the covered entity's information and the individual's agreement may be given orally.

- (a) Standard: uses and disclosures required by law.
 - (1) A covered entity may use or disclose protected health information to the extent that such use or disclosure is required by law and the use or disclosure complies with and is limited to the relevant requirements of such law.
 - (2) A covered entity must meet the requirements described in paragraph (c), (e), or (f) of this section for uses or disclosures required by law.
- (b) Standard: uses and disclosures for public health activities.
 - (1) Permitted disclosures. A covered entity may disclose protected health information for the public health activities and purposes described in this paragraph to:
 - (i) A public health authority that is authorized by law to collect or receive such information for the purpose of preventing or controlling disease, injury, or disability, including, but not limited to, the reporting of disease, injury, vital events such as birth or death, and the conduct of public health surveillance, public health investigations, and public health interventions;...

[Balance of the regulation section omitted. The reader is referred to the OCR website for further details: http://www.os.dhhs.gov/ocr/hipaa/.]

Chapter 3 Case Definition

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*This document may be reviewed or downloaded from the NBDPN website at: http://www.nbdpn.org/bdsurveillance.html

3.1 Introduction

A *case definition* is a set of criteria that define the parameters of what is included for quantitative description and analysis. In birth defects surveillance a *case* refers to an individual with characteristics that fit into the defined parameters. Important characteristics in birth defects surveillance include the diagnosis, pregnancy outcome information, and demographics.

In the absence of a single national birth defects surveillance program in the United States, pooled data from state-based programs across the country serve to estimate national rates, indicate regional variations, and describe the epidemiology of defects that occur rarely. Because, at any given time, these programs may be in different stages of development, employ different methods of ascertainment, and have different goals and objectives, the elements of the case definition used by each must be clearly identified in order to make valid comparisons and to minimize birth defects rate variations across surveillance programs and among individual defects ascertained by the same program.

Therefore, it is necessary for a surveillance program to develop a clear, concise case definition. Consistent application of a standard definition facilitates the accurate monitoring of clinically relevant conditions, identification of true changes over time, and comparison among populations in order to meet surveillance goals.

In the remainder of this chapter we discuss what is meant by the term 'birth defect' (Section 3.2), some important terminology for case definition (Section 3.3), and case definition criteria (Section 3.4). The relationship between case definition and the two terms 'sensitivity' and 'specificity' is discussed in Section 3.5. References cited in this chapter may be found in Section 3.6. Appendices to this chapter include birth defects included in the NBDPN's case definition (Appendix 3.1), the NBDPN Abstractor's Instructions (Appendix 3.2, available in electronic format at http://www.nbdpn.org/bdsurveillance.html), examples of minor anomalies (Appendix 3.3), and conditions related to prematurity (Appendix 3.4).

3.2 What Is Meant by a 'Birth Defect'

The general term 'birth defect' may take on a variety of meanings depending on the context in which it is used and the perspective of the person using it. 'Congenital abnormality', 'congenital anomaly', and 'congenital malformation' are terms often used as synonyms for 'birth defect'. However, the word 'congenital' may describe any condition present at birth, regardless of its etiology or timing of occurrence. In the broadest sense, the term *birth defect* encompasses a diversity of conditions including physical malformations, sensory deficits, chromosomal abnormalities, metabolic defects, neurodevelopmental disorders, and complications related to prematurity and low birth weight, among others.

While such a broad definition may be very helpful when seeking legislation and funding for screening, intervention, or prevention programs, a more specific definition is needed for surveillance purposes. Traditionally, birth defects surveillance programs have monitored major structural and genetic defects that adversely affect health and development (Correa-Villaseñor et al., 2003). The specific conditions monitored by an individual program will vary depending on the goals and objectives of that program, the case ascertainment methods used, and the resources available.

3.3 Terminology ¹

General Terminology

Major anomaly

A congenital abnormality that requires medical or surgical treatment, has a serious adverse effect on health and development, or has significant cosmetic impact. Individual major anomalies occur in less than 1 percent of the population. Together, they are seen in approximately 3 percent of births. Examples include cleft lip and tracheo-esophageal fistula.

Minor anomaly

A congenital abnormality that does not require medical or surgical treatment, does not seriously affect health and development, and does not have significant cosmetic impact. Individual minor anomalies generally occur in less than 4 percent of the population. The presence of multiple minor anomalies in the same child may provide clues to the timing of a prenatal insult and may indicate the presence of an undiagnosed major anomaly, syndrome, or functional deficit. Examples of minor anomalies are listed in Appendix 3.3.

Normal variant

A minor anomaly that occurs in approximately 4 percent or more of the population. Examples of normal variants include webbing of the second and third toes and a single umbilical artery in an otherwise normal infant.

Terminology Related to the Formation of Major Anomalies

Malformation

A major anomaly that arises during the initial formation of a structure, i.e., during organogenesis. For most organs, this occurs during the first eight weeks after fertilization. The resulting structure may be abnormally formed, incompletely formed, or may fail to form altogether. Examples of malformations include spina bifida and hypoplastic left heart. The term 'congenital malformation' is also used more broadly to indicate any major anomaly.

Disruption

A major anomaly that results from alteration of a structure after its initial formation. The resulting structure may have an altered shape and configuration, abnormal division or fusion of its component parts, or loss of parts that were previously present. Examples of disruption defects include intestinal atresia and possibly gastroschisis.

Deformation

A major anomaly that results from molding of part of a structure, usually over a prolonged time, by mechanical forces after its initial formation. Examples of forces that may lead to a deformation include oligohydramnios (diminished amniotic fluid) and intrauterine crowding in twin, triplet, or higher order pregnancies. Examples of deformations include the compression (Potter's) facies seen with bilateral renal agenesis and some instances of clubfoot.

¹ Stevenson, et al., 1993; Jones, 1997; Cunningham et al., 2001; Moore, 1977; National Center for Health Statistics, 2002.

Terminology Related to Patterns of Multiple Anomalies Occurring in a Single Child

Syndrome A pattern of anomalies that form a specific diagnosis for which the natural

history and recurrence risk are usually known. Use of the term 'syndrome' implies that the anomalies have a common specific etiology. Examples include Beckwith-Weidemann syndrome and Rubinstein-Taybi syndrome.

Sequence A pattern of anomalies that results from a single primary anomaly or

mechanical factor. The presence of the initial anomaly or factor leads to one or more secondary anomalies, which may then lead to one or more tertiary anomalies, etc., in cascade fashion. Examples include Robin sequence (micrognathia, posterior displacement of the tongue, cleft soft palate) and the oligohydramnios, or Potter, sequence (pulmonary hypoplasia, flattened facies,

abnormal positioning of the limbs).

Association A nonrandom pattern of anomalies that occur together more frequently than

expected by chance alone, but for which no etiology has been demonstrated. Examples include *VACTERL* association (Vertebral, Anal, Cardiac, Tracheo-

Esophageal, Renal, and Limb anomalies) and *CHARGE* association (Colobomas, Heart defects, choanal Atresia, Retarded growth and development and/or central nervous system anomalies, Genital anomalies and/or hypogondaism, Ear anomalies and/or deafness). Use of the term 'association' does not indicate that a specific diagnosis has been made.

Terminology Related to Tissue and Organ Formation

Agenesis Failure of an organ to form.

Dysgenesis Anomalous or disorganized formation of an organ.

Aplasia Absence of a tissue or organ due to lack of cell proliferation.

Dysplasia Disorganized cell structure or arrangement within a tissue or organ.

Hypoplasia Undergrowth of a tissue or organ due to insufficient proliferation of otherwise

normal cells.

Hyperplasia Overgrowth of a tissue or organ due to excess proliferation of otherwise

normal cells.

Terminology Related to the Timing of Gestation and Delivery

Embryonic period The first eight weeks after fertilization, during which most, but not all,

organs are formed.

Fetal period The period from the ninth week after fertilization through delivery.

Neonatal (newborn)

period

The first 28 days following delivery of a live-born infant.

T	D-1-4-3 4-41	Timing of Gestation	J D -1!	(491)
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Prenatal Before delivery.

Perinatal Before, during, or after delivery. The exact time period may vary from 20 to

28 completed weeks of gestation through 7 to 28 days after delivery,

depending on the context in which the term is used.

Postnatal After delivery.

Terminology Related to Pregnancy Outcome

Live birth Spontaneous delivery of an infant that exhibits signs of life, including a

> heartbeat, spontaneous breathing, or movement of voluntary muscles. Transient cardiac contractions and fleeting respiratory efforts or gasps are

not necessarily considered signs of life by all programs.

Fetal death (stillbirth) Spontaneous delivery of an infant or fetus at 20 weeks or greater gestation

> that does not exhibit signs of life. Transient cardiac contractions and fleeting respiratory efforts or gasps are not necessarily considered signs of life by all programs. A late fetal death is a fetal death that occurs at 28

The purposeful interruption of pregnancy with the intention other than to

weeks or greater gestation.

Spontaneous abortion

(miscarriage)

Spontaneous delivery of a fetus at less than 20 weeks gestation.

Induced abortion (elective termination)

produce a live birth and which does not result in a live birth.

Term infant An infant born after 37 completed weeks and before 42 completed weeks of

gestation.

Preterm infant An infant born before 37 completed weeks of gestation.

An infant born after 42 completed weeks of gestation. Postterm infant

Low birth weight Birth weight less than 2,500 grams, regardless of gestational age.

Very low birth weight Birth weight less than 1,500 grams, regardless of gestational age.

Extremely low birth

weight

Birth weight less than 1,000 grams, regardless of gestational age.

Neonatal death Death of a live-born infant within the first 28 days after birth. *Early*

neonatal death refers to death during the first 7 days. Late neonatal death

refers to death after 7 days but before 29 days.

Infant death Death of a live-born infant before 12 months of age.

3.4 Case Definition Criteria

In this section we discuss the various components of the case definition, that is, the criteria a birth defects surveillance program uses to define a case. These include diagnoses to be included (Section 3.4.1), residence (Section 3.4.2), pregnancy outcome (Section 3.4.3), gestational age (Section 3.4.4), age at which defects are diagnosed (Section 3.4.5), as well as the issue of pregnancies resulting from assisted reproductive technology (Section 3.4.6). Each of these criteria is discussed further below.

3.4.1 Diagnoses to Be Included

For the purposes of generating and reporting birth defects surveillance data across multiple states, the National Birth Defects Prevention Network (NBDPN) recommends the 45 major anomalies listed in Appendix 3.1. These were chosen on the basis of their frequency, their impact on public health, the state of knowledge about their etiologies and risk factors, and other considerations. Individual surveillance programs may want to expand this list to include additional defects of interest. Programs with limited resources may need to ascertain a subset of this list. Descriptions of each of the 45 diagnoses, its ICD-9-CM and CDC/BPA codes, inclusion and exclusion criteria, and defect-specific information that may be helpful when abstracting medical records are provided in the NBDPN Abstractor's Instructions posted on the NBDPN website (see Appendix 3.2). Examples of conditions considered to be minor anomalies are provided in Appendix 3.3. Conditions related to prematurity that are not considered to be major anomalies are listed in Appendix 3.4.

3.4.2 Residence

When monitoring the frequency of any condition, it is critical to define the population in which the cases occur. This allows one to calculate rates within the population, evaluate changes in these rates over time, plan for prevention and intervention services, and assess program goals and effectiveness. Population-based birth defects surveillance programs should strive to ascertain defects that occur among the offspring of all women who reside within a defined geographic area at the time of pregnancy outcome.

While this charge for surveillance programs appears straightforward, there are some special considerations. One such consideration is the fact that women who reside in one state or community may travel outside that area – such as to an adjacent state, specialty care center, or military facility – for obstetric care. In these instances, the mother's place of residence at the time of delivery (rather than the actual location of the delivery) should be used to determine whether to include her pregnancy in the surveillance. Including in-area residents who deliver outside the surveillance area, and excluding out-of-area residents who deliver within the surveillance area, is essential in order to conduct comprehensive surveillance. Whether an individual program attains this level of comprehensiveness will depend on how frequently women travel outside the surveillance area for delivery, the magnitude of the potential impact this may have on defect rates, the staff and resources available, and, most importantly, the existence of data-sharing relationships with facilities and programs outside the surveillance area. Recent changes in regulations concerning the privacy of medical records under the Health Information Portability and Accountability Act (HIPAA) have added to the complexity of these data-sharing relationships. The HIPAA regulations are discussed further in Chapter 2 on Legislation.

Another consideration is the fact that a surveillance program may identify more than one residential address for an individual woman. For example, the address of the health insurance policyholder listed in a hospital delivery record may differ from the mother's address listed on a birth certificate. If a patient changes residence during pregnancy, programs that employ multisource ascertainment may identify one

address from prenatal or laboratory records and another from the hospital delivery record. For these reasons, surveillance programs should develop standard procedures for deciding which of multiple addresses to accept as the mother's residence at delivery. Usually, this is the address at the time of delivery as listed on the vital record. If a vital record is not available or is not generated, as when an elective termination is performed outside the hospital setting, considering the mother's address from the termination record or from the prenatal visit closest in time to the delivery may be appropriate alternatives.

A third consideration requires detailing the method of determining whether a particular address lies within the surveillance area, particularly if the population under surveillance is not that of an entire state. For example, zip codes often cross city or county boundaries, streets may be renamed, new zip codes may be added, and city or county boundaries may change over time. Addresses that contain only a post office box number do not provide information about a person's actual place of residence. For these reasons, surveillance programs should develop standard procedures for distinguishing addresses that lie within or outside their surveillance area. Potential reference sources include street maps, United States Postal Service listings, tax assessor records, census tracts, and the latitude and longitude of the surveillance area (geocoding). While the latter can be extremely precise, the accuracy of geocoding will depend upon the accuracy of the addresses to which the latitude and longitude are assigned.

3.4.3 Pregnancy Outcome

Ideally, for births defects surveillance to be comprehensive with high sensitivity, all defects occurring in a population should be ascertained regardless of whether a pregnancy ends in live birth, fetal death, or spontaneous abortion, or whether an elective termination is performed. It is estimated that approximately 10 to 15 percent of all recognized pregnancies end in spontaneous abortion, and approximately 6 to 7 percent of those that reach 20 weeks gestation end in fetal death (Gabbe et al., 1996; National Center for Health Statistics, 2001). Surveillance systems that ascertain defects only among live-born infants may report incomplete data for defects that occur frequently among these outcomes. However, it is important to recognize that even late fetal deaths may not be scrutinized for defects as closely or as systematically as are live births. Unless an autopsy (including internal examination) and chromosome analysis are performed routinely, defects present in fetal deaths, yet not immediately evident in the delivery room, may remain unidentified. Even if an autopsy and chromosome analysis are performed, the presence of minor defects may not be recognized and syndromes may not be diagnosed. Whether it is beneficial for an individual program to ascertain defects reported in outcomes other than live births will depend upon the program's goals and objectives, the staff and resources available, the accessibility of information about these outcomes, and the magnitude of the potential impact on individual defect rates of excluding them.

The development and widespread use of prenatal diagnostic technology has posed additional issues for birth defects surveillance. These procedures have provided women with the option of electively terminating affected pregnancies, particularly those with defects that are life-threatening or that are likely to result in significant mental or functional impairment, usually before 20 weeks gestation. In the absence of prenatal diagnosis, many of these pregnancies would end in live birth or fetal death and would be included in birth defects surveillance data from many programs. Failure to ascertain prenatal diagnoses among electively terminated pregnancies may, therefore, limit the comprehensiveness and sensitivity of surveillance programs for some defects, such as neural tube defects and chromosomal abnormalities, even when defects among fetal deaths are ascertained. And – because the availability and utilization of prenatal diagnosis and elective termination may vary among populations, across geographic regions, and over time – the ability to make valid comparisons of some defect rates may be compromised unless pregnancies electively terminated after prenatal diagnosis are regularly ascertained.

Unfortunately, including these prenatal diagnoses will likely require expansion of a program's case ascertainment sources to include settings such as prenatal diagnostic clinics and termination centers. Furthermore, as is the case with fetal deaths, pregnancies that are electively terminated may not be fully scrutinized for confirmation of the prenatal diagnosis or the presence of additional defects or syndromes upon completion of the procedure. Again, whether it is beneficial for an individual program to ascertain defects reported in these pregnancies will depend upon the program's goals and objectives, the staff and resources available, the accessibility of information about these outcomes, and the magnitude of the potential impact on individual defect rates of excluding them. Regardless, it is important for birth defects surveillance programs to clearly state which outcomes are included when reporting surveillance data and to include pregnancies electively terminated after prenatal diagnosis whenever possible.

3.4.4 Gestational Age

Another important component of the case definition is the gestational age at delivery of the cases included in the surveillance data. The frequency of some defects may vary by gestational age, leading to variations in their rates depending on the length of gestation. For example, some defects are identified more frequently among preterm infants (Rasmussen et al., 2001; Shaw et al., 2001). Others, such as patent ductus arteriosus and undescended testes, may be abnormal in term infants but physiologically normal in preterm infants. Some ventricular septal defects that are present at birth in preterm infants might have closed during the last weeks of gestation if the pregnancy had continued to term. If surveillance systems differ in the gestational age at delivery of cases they include, or in their use of exclusion criteria based on gestational age, their rates of some defects may not be comparable.

Again, the inclusion of pregnancies electively terminated after prenatal diagnosis poses additional issues. Many of these pregnancies would have delivered spontaneously at a considerably later gestational age had they not been terminated. In order not to underestimate the frequency of defects for which elective termination may be performed, pregnancies terminated after prenatal diagnosis should be included in surveillance data regardless of the gestational age at which they were terminated. However, this may slightly overestimate the frequency of some defects relative to their frequency in the absence of prenatal diagnosis. For example, the majority of pregnancies electively terminated before 20 weeks gestation would have otherwise continued beyond 20 weeks to be included in birth defects surveillance programs that monitor pregnancies of 20 weeks or greater. However, a small proportion might have ended in spontaneous abortion before 20 weeks and would not appropriately be included in data from these programs. While the frequency of spontaneous abortion for pregnancies with Down syndrome has been estimated for each week of gestation, the natural history of pregnancies with other defects has not been well described (Hook et al., 1995). The frequency of spontaneous abortion by gestational week probably varies depending on the defect. Unfortunately, this effect is likely to be greater the earlier in gestation that affected pregnancies are terminated.

For the purposes of generating and reporting birth defects surveillance data across multiple states, the NBDPN recommends monitoring defects among live births and fetal deaths of 20 weeks or greater and among pregnancies electively terminated after prenatal diagnosis at any gestational age. Gestational age may be derived in various ways based on the date of the last menstrual period, measurement of the fetus by prenatal ultrasound, or the newborn clinical exam. Because these methods may not be equally accurate and may yield conflicting results, an important consideration is which method to use to determine whether a case fulfills the gestational age criterion for inclusion in surveillance data (Alexander et al., 1990; Hall, 1990). The methods below are listed in descending order of their generally accepted accuracy for calculating gestational age:

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- > Prenatal ultrasound with a reported gestational age of less than 14 weeks
- > Date of the last menstrual period
- ➤ Prenatal ultrasound with a reported gestational age of 14 weeks or greater
- ➤ Clinical examination after delivery

When multiple estimates of gestational age are ascertained for an individual case, the NBDPN recommends that the value derived using the method highest on this list be used to determine case status. Regardless of which method is used, it is important for birth defects surveillance programs to clearly state the gestational ages of the cases included when reporting surveillance data.

3.4.5 Age at Which Defects Are Diagnosed

The age at which a defect is diagnosed is also an important component of the case definition. The frequency of some defects may vary depending on the age of the child at diagnosis. While defects that are visible in the delivery room or symptomatic shortly after birth may be ascertained by most surveillance systems with high sensitivity, some internal defects may not be apparent for weeks or months after birth. Examples include cardiac defects that do not produce overt cyanosis, such as many atrial or ventricular septal defects, many obstructive renal defects, and some instances of intestinal malrotation. In addition, some chromosomal abnormalities may not be diagnosed until a year or more after birth when developmental delay or behavioral symptoms prompt a more in-depth evaluation. The rates of such conditions reported by surveillance systems that ascertain defects only among infants in the newborn nursery may not be comparable with those from systems that ascertain defects among older infants and children.

For the purposes of generating and reporting birth defects surveillance data across multiple states, the NBDPN recommends monitoring defects among live-born infants up to one year of age. Whether an individual program is able to ascertain defects beyond the newborn period will depend on the accessibility of information from sources other than the newborn nursery and the availability of staff and resources to add these additional sources. Programs should regularly state the range of ages at diagnosis included when reporting surveillance data.

As with other case definition criteria, the inclusion of defects that are diagnosed prenatally poses additional issues. The sensitivity and specificity of fetal ultrasound may vary for different defects depending on the gestational age, the skill and experience of the technician, the presence of maternal obesity, and other factors. The sensitivity and specificity of fetal ultrasound also may differ from that of newborn ultrasound and other postnatal diagnostic procedures. In addition, some conditions identified at mid-gestation by prenatal ultrasound may resolve spontaneously before delivery. Examples include renal obstructions, such as pyelectasis and uretero-pelvic junction obstructions, choroid plexus cysts of the brain, and some ventricular septal defects. Even chorionic villus sampling (CVS) may yield placental cells that contain chromosomal mosaicism not actually present in the fetus. For these reasons, many abnormalities diagnosed or suspected prenatally must be evaluated postnatally to determine their true nature. When such postnatal assessment is not possible or the medical records are not available, decisions about whether to include these defects in the surveillance must be made individually based on the certainty and specificity of the prenatal diagnosis for each case. General abstractor's instructions for the inclusion and exclusion of prenatal diagnoses for the 45 defects reported by the NBDPN are provided in Appendix 3.2. When reporting surveillance data, it is important for birth defects surveillance programs to state clearly the ages at which the defects were diagnosed and whether prenatal diagnoses without postnatal confirmation are included.

3.4.6 Pregnancies Resulting from Assisted Reproductive Technology

The use of assisted reproductive technology raises unique issues for birth defects surveillance, particularly in pregnancies where the egg from one woman (the biological mother) is used to conceive, but the pregnancy is carried by another woman (the birth mother). In this instance, genetic characteristics of the biological mother will be transmitted to the infant, but the birth mother's environment and lifestyle during pregnancy may also affect the infant. This situation may become quite complex when examining etiologic factors for birth defects. However, for surveillance purposes, the NBDPN recommends that the person listed on the child's birth certificate should be mother of record.

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3.5 Case Definition and Sensitivity and Specificity

Use of a consistent case definition is critical when evaluating the sensitivity and specificity of surveillance data and the efficiency and utility of surveillance programs.

The *sensitivity* of a surveillance program is defined as the proportion of cases occurring within a population that the program ascertains. Factors that may affect the sensitivity of a birth defects surveillance program include which pregnancy outcomes are ascertained (live births, fetal deaths, elective terminations), the gestational age at which they are ascertained (term infants only, pregnancies ≥ 20 weeks, all pregnancies), the child's age at the time the defect is diagnosed (prenatally, in the newborn period, before one year, at any age), and the diagnostic setting and methods used for ascertainment. For example, defects that are symptomatic in a live born infant may not be recognized in pregnancies that end in fetal death unless an autopsy is performed. Defects that are not immediately life-threatening, such as many cardiac septal defects, may not be diagnosed until several weeks or months after birth. If managed solely in the outpatient setting, these defects may be missed entirely by hospital-based programs unless surgical correction is required.

The *specificity* of a surveillance program is defined as the proportion of cases within a population that are ascertained by the program and that truly have defects. Factors that affect the sensitivity of a birth defects surveillance program may also affect its specificity. For example, patent ductus arteriosus (PDA) may be entirely normal in a preterm or one-day-old term infant but distinctly abnormal in a three-month-old child. Inclusion of all occurrences of PDA, regardless of gestational or postnatal age, may lead to ascertainment of false positive cases. Variations in the quality of prenatal ultrasound and in the natural course of some prenatal conditions necessitate postnatal confirmation of many diagnoses to avoid including false positive or clinically nonsignificant cases. Such confirmation may not be possible for pregnancies that end in fetal death or elective termination unless fetal autopsies are performed. Similarly, the exact nature of a congenital heart defect may not be finalized until the time of corrective surgery.

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3.6 References

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Appendix 3.1

Birth Defects Included in the Case Definition of the National Birth Defects Prevention Network

Appendix 3.1 Birth Defects Included in the Case Definition of the National Birth Defects Prevention Network

Birth Defects	n Defects ICD-9-CM Codes			
Birth Defects ICD-9-CM Codes CDC/BPA Codes Central Nervous System				
Anencephalus	740.0 - 740.1	740.00 - 740.10		
Spina bifida without anencephalus	741.0, 741.9	741.00 - 741.99		
	w/o 740.0 -740.10	w/o 740.0 - 740.10		
Hydrocephalus without Spina Bifida	742.3 w/o 741.0, 741.9	742.30 - 742.39		
		w/o 741.00 - 741.99		
Encephalocele	742.0	742.00 - 742.09		
Microcephalus	742.1	742.10		
	Eye			
Anophthalmia/microphthalmia	743.0, 743.1	743.00 - 743.10		
Congenital cataract	743.30 - 743.34	743.32 - 743.326		
Aniridia	743.45	743.42		
	Ear			
Anotia/microtia	744.01, 744.23	744.01, 744.21		
	Cardiovascular			
Common truncus	745.0	745.00 - 745.01		
Transposition of great arteries	745.10, .11, .12, .19	745.10 - 745.19		
Tetralogy of Fallot	745.2	745.20 - 745.21, 746.84		
Ventricular septal defect	745.4	745.40 - 745.490 (exclude 745.498)		
Atrial septal defect	745.5	745.50 - 745.59 (exclude 745.50)		
Endocardial cushion defect	745.60, .61, .69	745.60 - 745.69		
Pulmonary valve atresia and stenosis	746.01, 746.02	746.00 - 746.01		
Tricuspid valve atresia and stenosis	746.1	746.10		
•		(exclude 746.105)		
Ebstein's anomaly	746.2	746.20		
Aortic valve stenosis	746.3	746.30		
Hypoplastic left heart syndrome	746.7	746.70		
Patent ductus arteriosus (Include only if weight=>2500 grams or note if unable to exclude <2500 grams infants.)	747.0	747.00		
Coarctation of aorta	747.10	747.10 - 747.19		
	Orofacial			
Cleft palate without cleft lip	749.0	749.00 - 749.09		
Cleft lip with and without cleft palate	749.1, 749.2	749.10 - 749.29		
Choanal atresia 748.0 748.00		748.00		

Gastrointestinal				
Birth Defects	ICD-9-CM Codes	CDC/BPA Codes		
Esophageal atresia/tracheoesophageal fistula	750.3	750.30 - 750.35		
Rectal and large intestinal atresia/stenosis	751.2	751.20 - 751.24		
Pyloric stenosis	750.5	750.51		
Hirshsprung's disease (congenital megacolon)	751.3	751.30 - 751.34		
Biliary atresia	751.61	751.65		
	Genitourinary			
Renal agenesis/hypoplasia	753.0	753.00 - 753.01		
Bladder exstrophy	753.5	753.50		
Obstructive genitourinary defect	753.2, 753.6	753.20-29 - 753.60- 69		
Hypospadias and Epispadias	752.61, 752.62	752.600 - 752.627		
		(excluding 752.621)		
	Musculoskeletal			
Reduction deformity, upper limbs	755.20 - 755.29	755.20 - 755.29		
Reduction deformity, lower limbs	755.30 - 755.39	755.30 - 755.39		
Gastroschisis	756.79	756.71		
Omphalocele	756.79	756.70		
Congenital hip dislocation	754.30, .31, .35	754.30		
Diaphragmatic hernia	756.6	756.610 - 756.617		
	Chromosomal			
Trisomy 13	758.1	758.10 - 758.19		
Down syndrome (Trisomy 21)	758.0	758.00 - 758.09		
Trisomy 18	758.2	758.20 - 758.290		
	Other			
Fetal alcohol syndrome	760.71	760.71		
Amniotic bands	No code	658.80		

Appendix 3.2 NBDPN Abstractor's Instructions

Appendix 3.2 NBDPN Abstractor's Instructions

Format for Birth Defect Descriptions	
Central Nervous System	
Anencephalus	A3.2-2
Spina bifida without anencephalus	A3.2-3
Hydrocephalus without Spina Bifida.	A3.2-5
Encephalocele	A3.2-7
Microcephalus	A3.2-8
Wilciocephatus	A3.2-0
Eye	
Anophthalmia/microphthalmia	A3.2-10
Congenital cataract	
Aniridia	
Ear	
Anotia/microtia.	A3.2-13
Cardiovascular	
Common truncus	
Transposition of great arteries	A3.2-16
Tetralogy of Fallot	
Ventricular septal defect	A3.2-19
Atrial septal defect	A3.2-20
Endocardial cushion defect	
Pulmonary valve atresia and stenosis	A3.2-23
Tricuspid valve atresia and stenosis	A3.2-24
Ebstein's anomaly	A3.2-25
Aortic valve stenosis	A3.2-26
Hypoplastic left heart syndrome	A3.2-27
Patent ductus arteriosus	A3.2-28
Coarctation of aorta	A3.2-30
Orofacial	
Cleft palate without cleft lip	A3.2-31
Cleft lip with and without cleft palate	
Choanal atresia	
Esophageal atresia/tracheoesophageal fistula	
Rectal and large intestinal atresia/stenosis.	A3.2-35
Pyloric stenosis	
Hirshsprung's disease (congenital megacolon)	A3.2-37
Dillomestania	12220

Appendix 3.2 Case Definition

Genitourinary	
Renal agenesis/hypoplasia	A3.2-40
Bladder exstrophy	
Obstructive genitourinary defect	
Hypospadias and Epispadias	
Musculoskeletal	
Reduction deformity, upper limbs	A3.2-48
Reduction deformity, lower limbs	A3.2-51
Gastroschisis	A3.2-54
Omphalocele	A3.2-56
Congenital hip dislocation	
Diaphragmatic hernia	
Chromosomal	
Trisomy 13	A3.2-60
Down syndrome (Trisomy 21)	
Trisomy 18	A3.2-64
Other	
Fetal alcohol syndrome	A3.2-66
Amniotic bands	A3 2-67

Appendix 3.2 Case Definition

Appendix 3.2 NBDPN Abstractor's Instructions

Format for Birth Defect Descriptions

Defect Name		
Description	Description of the defect.	
Inclusions	Other names or conditions that should be included in the code for the defect.	
Exclusions	Other names or conditions that should not be included in the code for the defect.	
ICD-9-CM Codes	Applicable ICD-9-CM codes for the defect.	
CDC/BPA Codes	Applicable CDC/BPA codes for the defect.	
Diagnostic Methods	Postnatal procedures by which the defect may be accurately and reliably diagnosed.	
Prenatal Diagnoses Not Confirmed Postnatally	Guidance on whether cases with only a prenatal diagnosis should be included in the defect code.	
Additional Information	Tips and useful information about the defect.	

Description Partial or complete absence of the brain and skull.

Inclusions Acrania – Absence of skull bones with some brain tissue

present.

Absent brain, with or without skull bones present.

Anencephaly Anencephaly

Craniorachischisis – Anencephaly continuous with an open posterior spinal defect with no meninges covering the neural

tissue.

Exclusions Encephalocele

Iniencephaly

Rachischisis – When used alone, this term refers only to the spinal defect and should be coded as spina bifida without

anencephalus.

ICD-9-CM Codes 740.0 – 740.1

CDC/BPA Codes 740.00 – 740.10

Diagnostic Methods Anencephalus is easily recognized on physical examination at

delivery.

Prenatal Diagnoses Not Confirmed Postnatally Anencephalus may be included when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance

data.

Additional Information:

Anencephalus is one of a group of defects that result from failure of the neural tube to close.

Maternal serum alphafetoprotein (MSAFP) and/or amniotic fluid alphafetoprotein (AFAFP) and amniotic fluid acetylcholinesterase (ACHE) may be elevated with anencephalus. However, these screening tests alone are not sufficient to diagnose the condition.

In cases where both anencephalus and spina bifida are present but are not continuous (i.e., not craniorachischisis), both anencephalus and spina bifida should be coded.

Spina Bifida without Anencephalus

Description Incomplete closure of the vertebral spine (usually posteriorly)

through which spinal cord tissue and/or the membranes covering

the spine (meninges) herniate.

Inclusions Lipomeningocele

Lipomyelomeningocele

Meningocele – Herniation of meninges only.

Meningomyelocele, Myelomeningocele – Herniation of

meninges and spinal cord tissue.

Myelocystocele Myelodysplasia Myeloschisis Open spina bifida

Rachischisis – Open spina bifida without meninges covering the

spinal cord tissue. Spina bifida aperta Spina bifida cystica

Exclusions Closed spina bifida

Diastematomyelia Diplomyelia Hydromyelia

Spina bifida with coexisting anencephalus – Code only as

anencephalus. Spina bifida occulta Syringomyelia Tethered spinal cord

ICD-9-CM Codes 741.0 or 741.9 without 740.0 – 740.1

CDC/BPA Codes 741.00 – 741.99 without 740.00 – 740.10

Diagnostic MethodsThe majority of defects result in a direct opening on the infant's

back that is easily recognized on physical examination at delivery. However, the exact nature of the defect (meningocele vs. myelomeningocele) may only be distinguished by CT or

MRI scan, at surgery, or at autopsy.

Prenatal Diagnoses Not

Spina bifida may be included when only diagnosed prenatally.

However, if it is possible to ascertain the degree of certainty of

the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. In addition, the absence of spina bifida on prenatal

ultrasound does not necessarily mean that it will not be

diagnosed after delivery.

Additional Information:

Spina bifida is one of a group of defects that result from failure of the neural tube to close.

Open lesions (spina bifida cystica, spina bifida aperta) are those with no covering or with only meninges covering the neural tissue. They usually leak cerebrospinal fluid. Closed lesions are covered by normal skin.

Closed lesions, or spina bifida occulta, do not produce an opening in the infant's back and may result only in a defect of the vertebral spine without significant herniation of neural tissue or neurologic impairment. When asymptomatic, it may be detected as an incidental finding on an x-ray or other test performed for a different indication.

Hydrocephalus and Arnold-Chiari malformation of the brain frequently, though not always, result from spina bifida. When present, there is no need to code them separately from the spina bifida.

Maternal serum alphafetoprotein (MSAFP) and/or amniotic fluid alphafetoprotein (AFAFP) and amniotic fluid acetylcholinesterase (ACHE) may be elevated in spina bifida. However, these screening tests alone are not sufficient to diagnose the condition.

In cases where both anencephalus and spina bifida are present but are not continuous (i.e., not craniorachischisis), both anencephalus and spina bifida should be coded.

If the defect coding system includes unique codes for different levels of spina bifida (cervical; thoracic; lumbar; sacral) and a defect involves more than one level (cervicothoracic; thoracolumbar; lumbosacral), the highest level at which it occurs should be coded (i.e., cervical; thoracic; lumbar). The highest level of involvement determines the degree of associated neurologic impairment.

Appendix 3.2 A3.2-4 Case Definition

Hydrocephalus without Spina Bifida

Description An increase in the amount of cerebrospinal fluid (CSF) within

the brain resulting in enlargement of the cerebral ventricles and

increased intracranial pressure.

Inclusions Aqueductal stenosis – Narrowing or incomplete patency of the

aqueduct of Sylvius between the third and fourth ventricles. This is the most common type of obstructive hydrocephalus (see

below).

Atresia of the foramina of Magendie and Luschka – Incomplete patency of the openings in the roof of the fourth ventricle

through which CSF normally flows out of the brain.

Communicating hydrocephalus – Impaired absorption of CSF, leading to an increased amount of CSF within the brain.

Dandy-Walker malformation

Hydranencephaly

Hydrocephalus, type not specified

Obstructive (noncommunicating) hydrocephalus – Obstruction

of the flow of CSF within or out of the brain.

Exclusions Hydrocephalus that results from a prior intracranial hemorrhage.

This may be seen particularly in preterm infants.

Hydrocephalus that occurs in association with spina bifida. Only

the appropriate spina bifida code should be used.

Ventriculomegaly

ICD-9-CM Codes 742.3 without 741.0 or 741.9

CDC/BPA Codes 742.30 – 742.39 without 741.00 – 741.99

Diagnostic Methods While severe cases may be suspected by physical examination at

delivery, hydrocephalus may be conclusively diagnosed only through direct visualization of the brain by cranial ultrasound, CT or MRI scan, surgery, or autopsy. While a child's head circumference may be increased for age, this measurement alone

is not sufficient to make the diagnosis.

Prenatal Diagnoses Not

Confirmed Postnatally

While hydrocephalus may be identified by prenatal ultrasound, it generally should not be included in surveillance data without

postnatal confirmation. However, if it is possible to ascertain the degree of certainty of the diagnosis on prenatal ultrasound, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Severe cases may be

included without postnatal confirmation. Live-born children who survive should always have confirmation of the defect postnatally before being included.

Additional Information:

Hydrocephalus has a variety of etiologies, including infection, hemorrhage, and tumors, as well as anatomic lesions of the brain such as agenesis of the corpus callosum, encephalocele, cysts, and some bone dysplasias. In many cases, the etiology is not known.

In its true form, Dandy-Walker malformation is a malformation of the cerebellum and not a form of hydrocephalus. However, the term Dandy-Walker variant has been used to denote atresia of the foramina of Magendie and Luschka, dilatation of the cisterna magna (the space between the cerebellum and the brainstem), or cerebellar cysts, all of which have the appearance of increased fluid in the posterior fossa of the brain. It is, somewhat incorrectly, included in the defect codes for hydrocepahlus.

In hydranencephaly, the cerebral hemispheres are largely replaced by fluid-filled sacs within a normal skull. Hydranencephaly is not a true form of hydrocephalus. It is, somewhat incorrectly, included in the defect codes for hydrocepahlus.

Ventriculomegaly refers to enlargement of the cerebral ventricles, as measured by ultrasound (either prenatal or postnatal), CT or MRI scan. The distinction between hydrocephalus and ventriculomegaly has not been clearly defined, and these terms may be used interchangeably. Ventriculomegaly may be described as mild, moderate, or severe. How these designations correlate with the presence of true hydrocephalus, particularly when seen on prenatal ultrasound, also has not been clearly defined.

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Description Herniation of brain tissue and/or meninges through a defect in

the skull. The hernia sac is usually covered by skin.

Inclusions Cephalocele

Cranial meningocele – Herniation of meninges only.

Encephalocele

Encephalomyelocele - Herniation through a defect in a portion

of both the skull and the upper spine.

Encephalocystomeningocele

Hydranencephalocele Meningoencephalocele

Ventriculocele

Exclusions NA

ICD-9-CM Codes 742.0

CDC/BPA Codes 742.00 – 742.09

Diagnostic MethodsMost cases of encephalocele are recognizable on physical

examination after delivery. However, they may be conclusively diagnosed only through direct visualization of the brain by cranial ultrasound, CT or MRI scan, surgery, or autopsy. This is particularly true for internal herniations through the sphenoid, maxillary, or ethmoid bones, the orbit, or pharynx.

Prenatal Diagnoses Not Confirmed Postnatally

Encephalocele may be included when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. In addition, the absence of a small encephalocele on prenatal ultrasound does not necessarily mean

that it will not be diagnosed after delivery.

Additional Information:

Encephaloceles are often included as one of a group of defects that result from failure of the neural tube to close.

Maternal serum alphafetoprotein (MSAFP) and/or amniotic fluid alphafetoprotein (AFAFP) and amniotic fluid acetylcholinesterase (ACHE) may be elevated with encephaloceles. However, these screening tests alone are not sufficient to diagnose the condition.

Occipital encephalocele is a component of Meckel-Gruber syndrome.

Microcephalus		
Description	A cranial vault that is smaller than normal for age. The size of the cranial vault is an indicator of the size of the underlying brain.	
Inclusions	Microcephalus Microcephaly Primary or True Microcephalus	
Exclusions	Microcephalus that is secondary to a birth or delivery complication or to a postnatal insult or trauma.	
ICD-9-CM Codes	742.1	
CDC/BPA Codes	742.10	
Diagnostic Methods	Microcephaly is usually easily diagnosed on physical examination by measurement of the occipitofrontal circumference (OFC, head circumference). However, there is difference of opinion as to what the lower limit of a normal head circumference should be (see below). Cranial ultrasound, CT or MRI scans may also reflect the diagnosis and contribute to the diagnosis of any underlying brain abnormalities.	
Prenatal Diagnoses Not Confirmed Postnatally	While this condition may be identified by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of microcephalus on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.	

Additional Information:

Microcephalus may be defined variously as an OFC less than the 10th, 5th, or 3rd percentile, or less than 2 or 3 standard deviations below the mean for age. There is no single accepted standard. Reference graphs differ in terms of the cut-off values displayed and the reference population used. Reference graphs for postnatal OFC growth usually are displayed separately for males and females.

In addition, it must be recognized that a proportion of normal children will have an OFC below any single cut-off value (i.e., 5% of the population has an OFC below the 5th percentile by definition). For this reason, only children who have been given a clinical diagnosis of microcephalus should be included in birth defects surveillance data. The diagnosis should not be assigned based on the OFC measurement at birth without corroborating evidence from the medical record that the child carries the diagnosis of microcephalus.

Microcephalus itself is not a primary malformation, but a sign that the brain is small. It has a wide variety of causes. It is a component of a number of genetic syndromes. It also may result from a primary brain abnormality or a prenatal, perinatal, or postnatal insult. Examples of the latter include intrauterine infection, such as rubella or cytomegalovirus (CMV); *in utero* exposure to

alcohol and some medications, such as isotretinoin or dilantin; hypoxia during delivery; chronic hypoxia complicating prematurity; postnatal meningitis; head trauma. Only cases of microcephalus that have onset before delivery should be included in surveillance data. Unfortunately, the timing of onset and the etiology often are not known.

Appendix 3.2 A3.2-9 Case Definition

Anophthalmia/Microphthalmia

Description Anophthalmia – Total absence of eye tissue or apparent absence

of the globe in an otherwise normal orbit.

Microphthalmia – Reduced volume of the eye. The corneal

diameter is usually less than 10 millimeters, or the

anteroposterior globe diameter is less than 20 millimeters.

Inclusions Anophthalmia

Microphthalmia

Nanophthalmia – Microphthalmia with normal internal eye (intraocular) structures. This is a distinct genetic condition.

Exclusions Small eyes or small palpebral fissures for which the diagnosis of

microphthalmia or anophthalmia has not been made.

Microcornea with otherwise normal eye size.

ICD-9-CM Codes 743.0, 743.1

CDC/BPA Codes 743.00 – 743.10

Diagnostic Methods These conditions are usually recognized on physical

examination after delivery, especially by an ophthalmologist. However, the anteroposterior diameter of the globe may be measured only by ultrasound, CT or MRI scan, or at autopsy.

Prenatal Diagnoses Not Confirmed Postnatally While these conditions may be identified by prenatal ultrasound,

they should not be included in surveillance data without

postnatal confirmation. In addition, the absence of anophthalmia or microphthalmia on prenatal ultrasound does not necessarily

mean that it will not be diagnosed after delivery.

Additional Information:

Microphthalmia may occur in association with colobomas (gaps) in the uvea, iris, choroid and/or optic nerve (colobomatous microphthalmia).

Anophthalmia and microphthalmia often are accompanied by malformations of the brain and face, and frequently are components of genetic syndromes.

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('And	Tenital	Cataract
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Description An opacity of the lens of the eye that has its origin prenatally.

Inclusions Anterior polar cataract

Cataract, type not specified

Infantile cataract Lamellar cataract Nuclear cataract

Posterior lentiglobus/lenticonus cataract

Posterior cortical cataract

Sectoral cataract Zonular cataract

Exclusions Any of the above types of cataract that has its origin after birth

Corneal opacities

ICD-9-CM Codes 743.30 – 743.34

CDC/BPA Codes 743.320 – 743.326

Diagnostic Methods Some cataracts are readily apparent on physical examination.

Others are visible with an ophthalmoscope. However, they may be conclusively diagnosed only through examination by an

ophthalmologist.

Prenatal Diagnoses Not Confirmed Postnatally While this condition may be identified by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of a cataract on prenatal ultrasound does not necessarily mean that it will not be

diagnosed after delivery.

Additional Information:

Cataracts may be congenital, acquired, or inherited. They may involve all or only part of the lens of either or both eyes. They may be an isolated finding in an otherwise normal eye, or may be part of a more general eye malformation. They may be seen with metabolic disorders, such as galactosemia; genetic syndromes, such as chondrodysplasia punctata; chromosomal abnormalities, such as Trisomy 21; intrauterine infection, such as congenital rubella; or trauma.

In some instances, the severity of the cataract progresses over time. The need for surgical treatment depends on the degree of visual impairment.

When congenital cataract occurs with microphthalmia in the same infant, both conditions should be coded.

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Description Hypoplasia of the iris of both eyes.

Inclusions Aniridia

Hypoplasia of the iris

Exclusions Axenfeld-Rieger syndrome

Chandler syndrome Coloboma of the iris

Iris atrophy Peters anomaly Rieger syndrome

ICD-9-CM Codes 743.45

CDC/BPA Codes 743.42

Diagnostic Methods Aniridia may be apparent on physical examination. However, it

may be conclusively diagnosed only through examination by an

ophthalmologist.

Prenatal Diagnoses Not Confirmed Postnatally Aniridia should not be included in surveillance data unless

diagnosed postnatally.

Additional Information:

Aniridia is usually associated with other abnormalities of the eye, including a persistent pupillary membrane; displaced lens; glaucoma; corneal and retinal abnormalities; hypoplasia of the optic nerve. While there is often near-total absence of the iris, it is never completely absent. Aniridia may be a component of Peters anomaly (abnormal development of the cornea and anterior chamber of the eye).

Aniridia has been associated with Wilms tumor of the kidney and certain chromosomal abnormalities. It may be inherited or may occur sporadically.

When aniridia occurs with a cataract in the same infant, both conditions should be coded.

Anotia/Microtia		
Description	Anotia – Total absence of the external ear and canal.	
	Microtia – Malformation or hypoplasia of the external ear (auricle, pinna).	
Inclusions	Anotia Microtia	
Exclusions	Small ears that retain most of the overall structure of the normal auricle, including lop or cup ear defects. In these, the auditory meatus is usually patent and defects of the ossicular chain of the middle ear are infrequent. However, these defects are sometimes designated as Type I Microtia.	
	Isolated absence, atresia, stenosis or malformation of the ear canal with a normal external ear.	
	Congenital absence of the ear not diagnosed as anotia or microtia.	
ICD-9-CM Codes	744.01, 744.23	
CDC/BPA Codes	744.01, 744.21	
Diagnostic Methods	Anotia and microtia are usually easily recognized on physical examination after delivery. However, abnormalities of the middle and inner ear may be conclusively diagnosed only by CT or MRI scan, surgery, or autopsy.	
Prenatal Diagnoses Not Confirmed Postnatally	While these conditions may be identified by prenatal ultrasound, they should not be included in surveillance data without postnatal confirmation. In addition, the absence of anotia or microtia on prenatal ultrasound does not necessarily mean that they will not be diagnosed after delivery.	

Additional Information:

The spectrum of severity of microtia may range from a measurably small external ear with minimal structural abnormality to major structural alteration of the external ear with an absent or blindending canal. Following is the classification system of Meurman (modified from Marks):

Type I B – Generally small ears that retain most of the overall structure of the normal auricle. These should not be coded as microtia.

Type II B-A moderately severe anomaly with a longitudinal mass of cartilage with some resemblance to a pinna. The rudimentary auricle may be hook-shaped, have an S-shape, or the appearance of a question mark.

Type III B – The ear is a rudiment of soft tissue and the auricle has no resemblance to a normal pinna.

Type IV B – Complete absence of all external ear structures (anotia).

Abnormalities that may be associated with anotia/microtia include anomalies of the middle and/or inner ear, the mandible and face, and hearing loss.

Anotia/microtia may be a component of Goldenhar and other syndromes.

Appendix 3.2 A3.2-14 Case Definition

Common Truncus (Truncus Arteriosus or TA)

Description Failure of separation of the aorta and the pulmonary artery,

resulting in a single common arterial trunk carrying blood from

the heart to both the body and lungs.

Inclusions Common truncus

Truncus arteriosus (TA)

Exclusions Aorto-pulmonary window

ICD-9-CM Codes 745.0

CDC/BPA Codes 745.00 – 745.01

Diagnostic Methods While truncus defects may be suspected by clinical

presentation, they may be conclusively diagnosed only through

direct visualization of the heart by cardiac echo

(echocardiography), catheterization, surgery, or autopsy.

Prenatal Diagnoses Not Confirmed Postnatally These conditions may be included as cases when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally.

Additional Information:

A ventricular septal defect is often present in association with truncus defects and should be coded separately.

Truncus arteriosus is one of several abnormalities of the outflow tract of the heart known as conotruncal defects. Some infants with these defects have a deletion on the short arm of chromosome 22 (22q11 deletion). This deletion is diagnosed using fluorescent *in situ* hybridization (FISH) and will not necessarily be detected on a routine karyotype analysis.

Transposition of the Great Arteries (TGA)

Description Transposition of the aorta and the pulmonary artery such that

the aorta arises from the right ventricle (instead of the left) and the pulmonary artery arises from the left ventricle (instead of

the right).

Inclusions Complete transposition (d-TGA without a VSD)

Corrected transposition (1-TGA)

Incomplete transposition (d-TGA with a VSD)

Transposition of the Great Arteries (TGA), not otherwise

specified

Transposition of the Great Vessels (TGV)

Exclusions NA

ICD-9-CM Codes 745.10, 745.11, 745.12, 745.19

CDC/BPA Codes 745.10 – 745.19

Diagnostic Methods While transposition defects may be suspected by clinical

presentation, they may be conclusively diagnosed only through

direct visualization of the heart by cardiac echo

(echocardiography), catheterization, surgery, or autopsy.

Prenatal Diagnoses Not Confirmed Postnatally These conditions may be included as cases when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally.

Additional Information:

In order for a child with d-TGA to survive, a communication must be present between the pulmonary and systemic circulations to allow oxygenated blood from the lungs to reach the right ventricle for distribution to the rest of the body through the abnormally placed aorta. In most instances, this communication is through a ventricular septal defect (incomplete TGA). If a VSD is not present, oxygenated blood from the lungs is returned directly to the lungs without being distributed to the rest of the body (complete TGA).

If the defect coding system does not include unique codes to differentiate TGA with and without a VSD (complete vs. incomplete), the VSD should be coded separately when present.

l-TGA (corrected transposition) is a defect in which the ventricle on the right side of the heart has the anatomic appearance of the left ventricle, and the ventricle on the left side of the heart has the anatomic appearance of the right ventricle (ventricular inversion). The pulmonary artery arises from the anatomic left ventricle and the aorta arises from the anatomic right ventricle (hence the designation of transposition). Because blood from the ventricle on the right flows through the pulmonary artery, and that from the ventricle on the left flows through the aorta, circulation is

normal as long as there are no other defects.

Transposition of the great arteries is one of several abnormalities of the outflow tract of the heart known as conotruncal defects. Some infants with these defects have a deletion on the short arm of chromosome 22 (22q11 deletion). This deletion is diagnosed using fluorescent *in situ* hybridization (FISH) and will not necessarily be detected on a routine karyotype analysis.

Tetralogy of Fallot		
Description	The simultaneous presence of a ventricular septal defect (VSD), pulmonic stenosis, a malpositioned aorta that overrides the ventricular septum, and right ventricular hypertrophy.	
Inclusions	Pentalogy of Fallot – Tetralogy of Fallot with an associated inter-atrial communication, either a patent foramen ovale (PFO) or an atrial septal defect (ASD). Tetralogy of Fallot Tet TOF	
	Some coding systems may also include Trilogy of Fallot, or Fallot's Triad – the simultaneous presence of an atrial septal defect, pulmonic stenosis, and right ventricular hypertrophy.	
Exclusions	Simultaneous occurrence of a VSD and pulmonary stenosis that has TOF physiology but has not been diagnosed as Tetralogy of Fallot.	
ICD-9-CM Codes	745.2	
CDC/BPA Codes	745.20 – 745.21, 746.84	
Diagnostic Methods	While Tetralogy of Fallot may be suspected by clinical presentation, it may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy.	
Prenatal Diagnoses Not Confirmed Postnatally	These conditions may be included as cases when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally.	

Additional Information:

Children with Tetralogy of Fallot may experience episodes of cyanosis or hypoxia that result from shunting of unoxygenated blood across the VSD from the right to the left ventricle. Children who have a coexisting VSD and pulmonary stenosis, but do not have Tetralogy of Fallot, may experience similar episodes. Thus, the occurrence of cyanosis or hypoxia does not necessarily mean a child has been diagnosed with Tetralogy of Fallot.

Tetralogy of Fallot is one of several abnormalities of the outflow tract of the heart known as conotruncal defects. Some infants with these defects have a deletion on the short arm of chromosome 22 (22q11 deletion). This deletion is diagnosed using fluorescent *in situ* hybridization (FISH) and will not necessarily be detected on a routine karyotype analysis.

Ventricular Septal Defect (VSD)

Description An opening in the septum that separates the left and right

ventricles of the heart.

Inclusions Ventricular septal defect

VSD

Exclusions Ventricular septal defects that occur as part of Tetralogy of

Fallot or an endocardial cushion defect. Inflow-type,

subtricuspid, and canal-type VSDs are assumed to be part of an endocardial cushion defect and should not be coded separately .

ICD-9-CM Codes 745.4

CDC/BPA Codes 745.40 – 745.59, excluding 745.498

Diagnostic Methods Some isolated VSDs may be diagnosed on physical examination

and/or EKG without direct imaging of the heart. However, many VSDs may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography),

catheterization, surgery, or autopsy.

Prenatal Diagnoses Not Confirmed Postnatally

While VSDs may be identified by prenatal ultrasound, many close spontaneously before delivery. For this reason, VSDs that are diagnosed prenatally should not be included unless they have been confirmed postnatally. In addition, the absence of a VSD on prenatal ultrasound does not necessarily mean that a VSD will not be diagnosed after delivery, as it is not always possible to accurately visualize the entire ventricular septum by

prenatal ultrasound.

Additional Information:

VSDs may be of several types, depending on the location of the opening along the ventricular septum. The most common are:

Muscular

Membranous

Perimembranous

However, in many instances the type of VSD may not be specified in the medical record.

Many muscular, membranous and perimembranous VSDs may close spontaneously in the first weeks or months of life without treatment.

An aneurysm of the ventricular septum indicates a membranous or perimembranous VSD that is in the process of closing.

Atrial Septal Defect (ASD)

Description An opening in the septum that separates the left and right atria

of the heart.

Inclusions Atrial septal defect, type not specified

ASD

Secundum ASD (ASD 2 or ASD II)

ASD vs. PFO – In the first days of life, it may not be possible to distinguish whether the opening in the atrial septum is a true ASD or a patent foramen ovale that has not yet closed (see below). ASD vs. PFO should be included only if the exact

nature of the condition was never resolved.

Exclusions Atrioventricular septal defects (AVSD) – These are included

under endocardial cushion defects (see below).

Patent foramen ovale (PFO) – A PFO is normal *in utero* and frequently does not close until 24 to 48 hours after birth.

Primum ASD (1° ASD) – These are included under

endocardial cushion defects (see below).

ICD-9-CM Codes 745.5

CDC/BPA Codes 745.50 – 745.59, excluding 745.50

Diagnostic Methods Some isolated ASDs may be diagnosed based on physical

examination and/or EKG without direct imaging of the heart. However, many ASDs may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy.

Prenatal Diagnoses Not Confirmed Postnatally

While ASDs may be identified by prenatal ultrasound, they may close spontaneously before delivery. For this reason, ASDs that are diagnosed prenatally should not be included unless they have been confirmed postnatally. In addition, the absence of an ASD on prenatal ultrasound does not necessarily mean that an ASD will not be diagnosed after delivery, as it is not always possible to accurately visualize the entire atrial

septum by prenatal ultrasound.

Additional Information:

Secundum ASDs are usually located toward the middle of the atrial septum. Some close spontaneously without treatment.

Primum ASDs are located in the lower portion of the atrial septum, are etiologically related to endocardial cushion (AV canal) defects, and never close spontaneously.

Endocardial Cushion Defect

Description

A defect in both the lower portion of the atrial septum and the upper portion of the ventricular septum, producing a large opening (canal) in the central part of the heart. The adjacent parts of the mitral and tricuspid valves may also be abnormal, resulting in a single common atrioventricular valve. In extreme cases, virtually the entire atrial and ventricular septae may be missing.

Inclusions

Atrioventricular septal defect (AVSD) Common or complete atrioventricular (AV) canal Common atrioventricular (AV) orifices Endocardial cushion defect

Primum atrial septal defect (1° ASD) – A defect only in the lower portion of the atrial septum. While this does not also involve a defect in the upper portion of the ventricular septum, it is etiologically related to the more complete form. A cleft mitral valve is often present.

Common atrium – A very large primum ASD. Incomplete AV canal (incomplete endocardial cushion defect) – Same as a primum ASD.

Inflow-type, subtricuspid, or canal-type ventricular septal defect (VSDAVC) – A defect in the upper (inflow) portion of the ventricular septum. While this does not also involve a defect in the lower portion of the atrial septum, it is etiologically related to the more complete form.

Exclusions

Secundum ASDs that coexist with a VSD. In this instance, both the ASD and the VSD should be coded.

ICD-9-CM Codes

745.60, 745.61, 745.69

CDC/BPA Codes

745.60 - 745.69

Diagnostic Methods

While endocardial cushion defects may be suspected by clinical presentation, examination, and EKG changes, it may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy.

Prenatal Diagnoses Not Confirmed Postnatally

These conditions may be included as cases when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data, as it may be difficult to

distinguish this condition from other abnormalities of the cardiac septae prenatally. Live-born children who survive should always have confirmation of the defect postnatally.

Additional Information:

Endocardial cushion defects are known to be associated with Down syndrome. Approximately 20% of children with Down syndrome have some type of endocardial cushion defect. Conversely, approximately 70% of children with an endocardial cushion defect have Down syndrome.

Pulmonary Valve Atresia and Stenosis

Description Pulmonary valve atresia – Lack of patency, or failure of

formation altogether, of the pulmonary valve, resulting in obstruction of blood flow from the right ventricle to the

pulmonary artery.

Pulmonary valve stenosis – Obstruction or narrowing of the pulmonary valve, which may impair blood flow from the right

ventricle to the pulmonary artery.

Inclusions Pulmonary valve atresia

Pulmonary valve stenosis Pulmonic stenosis (PS)

Exclusions Atresia or stenosis of the main or branch (right or left)

pulmonary arteries, not involving the pulmonary valve. Pulmonary stenosis that occurs as part of Tetralogy or

Pentalogy of Fallot.

Supra-valvular or sub-valvular pulmonic stenosis.

ICD-9-CM Codes 746.01, 746.02

CDC/BPA Codes 746.00 – 746.01

Diagnostic Methods While pulmonary valve atresia or stenosis may be suspected by

clinical presentation, it may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy.

Prenatal Diagnoses Not Confirmed Postnatally While these conditions may be identified by prenatal ultrasound, they should not be included in surveillance data without postnatal confirmation. In addition, the absence of pulmonary valve atresia or stenosis on prenatal ultrasound does not necessarily mean that it will not be diagnosed after

delivery.

Additional Information:

Pulmonary valve atresia or stenosis may occur with or without a coexisting ventricular septal defect. When it occurs with a VSD, the child may experience episodes of cyanosis or hypoxia similar to those seen in children with Tetralogy of Fallot. This results from shunting of unoxygenated blood across the VSD from the right to the left ventricle. Thus, the occurrence of cyanosis or hypoxia does not necessarily mean that the child has Tetralogy of Fallot.

Tricuspid Valve Atresia and Stenosis

Description Tricuspid valve atresia – Lack of patency, or failure of

formation altogether, of the tricuspid valve, resulting in obstruction of blood flow from the right atrium to the right

ventricle.

Tricuspid valve stenosis – Obstruction or narrowing of the tricuspid valve, which may impair blood flow from the right

atrium to the right ventricle.

Inclusions Tricuspid atresia

Tricuspid stenosis

Exclusions Tricuspid regurgitation without specific mention of tricuspid

atresia or stenosis.

ICD-9-CM Codes 746.1

CDC/BPA Codes 746.10 (excluding 746.105)

Diagnostic Methods While tricuspid valve atresia or stenosis may be suspected by

clinical presentation, it may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy.

Prenatal Diagnoses Not Confirmed Postnatally While these conditions may be identified by prenatal ultrasound, they should not be included in surveillance data without postnatal confirmation. In addition, the absence of tricuspid valve atresia or stenosis on prenatal ultrasound does

not necessarily mean that it will not be diagnosed after

delivery.

Additional Information NA

Ebstein's Anomaly	
Description	Downward displacement if the tricuspid valve into the right ventricle. The tricuspid valve is usually hypoplastic and regurgitant.
Inclusions	Ebstein's anomaly Ebstein malformation
Exclusions	NA
ICD-9-CM Codes	746.2
CDC/BPA Codes	746.20
Diagnostic Methods	While Ebstein's anomaly may be suspected by clinical presentation, it may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy.
Prenatal Diagnoses Not Confirmed Postnatally	While this condition may be identified by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of Ebstein's anomaly on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.

Additional Information:

Ebstein's anomaly has been associated with lithium exposure during gestation. However, the magnitude of this association is probably very small.

	Aortic Valve Stenosis
Description	Obstruction or narrowing of the aortic valve, which may impair blood flow from the left ventricle to the aorta.
Inclusions	Stenosis of the aortic valve
Exclusions	Stenosis of the aorta without mention of the aortic valve. Supra-valvular or sub-valvular aortic stenosis.
ICD-9-CM Codes	746.3
CDC/BPA Codes	746.30
Diagnostic Methods	While aortic valve stenosis may be suspected by clinical presentation, it may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy.
Prenatal Diagnoses Not Confirmed Postnatally	While this condition may be identified by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of aortic valve stenosis on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.
Additional Information	NA

Hypoplastic Left Heart Syndrome (HLHS)

Description A condition in which the structures on the left side of the heart

and the aorta are extremely small. Classically, this condition includes hypoplasia of the left ventricle, atresia or severe hypoplasia of the mitral and aortic valves, and hypoplasia and

coarctation of the aorta.

Inclusions Any diagnosis of hypoplastic left heart syndrome, regardless of

whether all conditions in the classical definition are present.

Exclusions Hypoplasia or diminished size of the left ventricle alone

without involvement of other structures on the left side of the

heart or the aorta.

Hypoplastic left heart or small left ventricle that occurs as part

of another complex heart defect, such as an endocardial

cushion defect (AV canal).

ICD-9-CM Codes 746.7

CDC/BPA Codes 746.70

Diagnostic Methods While hypoplastic left heart may be suspected by clinical

presentation, examination, and EKG changes, it may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization,

surgery, or autopsy.

Prenatal Diagnoses NotThese conditions may be included as cases when only

Confirmed Postnatally diagnosed prenatally. However, if it is possible to ascertain the

degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual

case in the surveillance data, as it may be difficult to

distinguish this condition from other abnormalities of the left ventricle prenatally. Live-born children who survive should always have confirmation of the defect postnatally before being

included.

Additional Information NA

Patent Ductus Arteriosus (PDA)

Description Abnormally persistent blood flow through the ductus arteriosus

beyond the first few days of life.

Inclusions Patent ductus arteriosus in infants with birth weight $\geq 2,500$

grams who have not been given prostaglandin (see below).

Exclusions Patent ductus arteriosus in infants with birth weight < 2,500

grams.

Patent ductus arteriosus in infants $\geq 2,500$ grams who have

been given prostaglandin (see below).

ICD-9-CM Codes 747.0

CDC/BPA Codes 747.00

Diagnostic Methods Some instances of patent ductus arteriosus may be diagnosed

on physical examination. However, many PDAs may be diagnosed conclusively only through direct visualization of the heart by cardiac echo (echocardiography), catheterization,

surgery, or autopsy.

Prenatal Diagnoses Not Confirmed Postnatally Because a patent ductus arteriosus is normal and necessary during fetal life, this condition should not be included in surveillance data unless diagnosed postnatally at an appropriate

age.

Additional Information:

In the normal fetal circulation, blood flows from the right ventricle to the pulmonary artery, then crosses the ductus arteriosus to the aorta for distribution to the body and the placenta. This bypasses much of the pulmonary circulation, since fetal blood is oxygenated by the placenta and not the lungs. Over the first hours after a normal full-term birth, smooth muscle in the wall of the ductus contracts and thickens to prevent blood flow through the ductus. Over the subsequent 2 to 3 weeks of life, the ductus is replaced by fibrous tissue and the communication is permanently sealed. Persistence of a patent ductus through which blood may flow beyond that time is abnormal.

In preterm infants, the ability of the ductus to constrict and close after delivery is not fully developed. Patent ductus arteriosus in a preterm infant is more likely to be a consequence of prematurity rather than an inherent abnormality. In these infants, it should not be coded as a defect.

The length of time required for the ductus to close is somewhat variable among term infants, and there is disagreement among specialists about the length of time after which patency is abnormal. Some birth defects surveillance programs only include PDAs that have been present for at least 6 weeks after birth

Term infants who have additional heart defects may have abnormal patterns of blood flow or abnormal pressures in the pulmonary artery and aorta which prevent the ductus from closing. In these instances, the PDA is not an inherent abnormality but secondary to the additional defects.

In some severe heart defects, such as pulmonary atresia or d-TGA without a VSD, the infant's initial survival may depend on the presence of a patent ductus arteriosus in order for blood to reach the lungs for oxygenation. Prostaglandin (PGE) may be administered intravenously to maintain the patency of the ductus. In these instances, the PDA is an artifact of treatment of the underlying condition and should not be coded as a defect.

Patent ductus arteriosus may be a component of persistent transitional (fetal) circulation, in which the fetal pattern of blood flow through the ductus and bypassing the lungs, persists after birth. This is often a physiologic response to hypoxia from respiratory suppression, as may be seen with meconium aspiration.

	Coarctation of the Aorta
Description	Narrowing of the descending aorta, which may obstruct blood flow from the heart to the rest of the body. The most common site of coarctation occurs distal to the origin of the left subclavian artery in the region of the ductus arteriosus.
Inclusions	Coarctation of the aorta, type not specified Preductal, juxtaductal, and postductal coarctations – These terms refer to the exact placement of the segment of coarctation relative to the insertion of the ductus arteriosus.
Exclusions	NA
ICD-9-CM Codes	747.10
CDC/BPA Codes	747.10 – 747.19
Diagnostic Methods	While coarctation of the aorta may be suspected by clinical presentation and examination, it may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy.
Prenatal Diagnoses Not Confirmed Postnatally	While this condition may be identified by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of coarctation of the aorta on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.

Additional Information:

Left-sided obstructive lesions of the heart, such as coarctation, have been associated with Turner syndrome (karyotype 45,X and other variants).

Cleft Palate without Cleft Lip

Description An opening in the roof of the mouth resulting from incomplete

fusion of the shelves of the palate. The opening may involve

the hard palate only, the soft palate only, or both.

Inclusions Bifid or cleft uvula

Cleft palate, type not specified

Cleft hard palate Cleft soft palate

Submucous cleft palate – A cleft in the soft palate that is

covered by the mucosa or a thin muscle layer.

Exclusions Cleft palate that coexists with a cleft lip. These should be

coded as cleft lip only (see below).

ICD-9-CM Codes 749.0

CDC/BPA Codes 749.00 – 749.09

Diagnostic Methods Cleft palate is usually easily recognized on physical

examination by direct visualization of the pharynx after delivery. It may also be seen on CT or MRI scan, at surgery or autopsy. However, submucous cleft palate may be difficult to diagnose by physical examination during the first year of life.

Prenatal Diagnoses Not Confirmed Postnatally While this condition may be identified by prenatal ultrasound, it should not be included in surveillance data without postnatal

confirmation. In addition, the absence of cleft palate on

prenatal ultrasound does not necessarily mean that it will not be

diagnosed after delivery.

Additional Information:

Cleft palate may be unilateral, bilateral, or central in location. If the defect coding system includes unique codes for these different types, the location of the cleft should be coded.

Cleft palate sometimes may be described as U-shaped or V-shaped. This distinction is not clinically meaningful and these conditions should not be coded differently.

Bifid uvula is often seen in association with a submucous cleft palate. However, bifid uvula also may occur alone. The presence of submucous cleft palate does not necessarily mean that a bifid uvula is present.

Cleft palate is one component of the Pierre Robin sequence, which also includes micrognathia and glossoptosis (when the tongue falls backward into the posterior pharynx). When diagnosed, Pierre Robin sequence should be coded separately.

Cleft Lip with and without Cleft Palate

Description A defect in the upper lip resulting from incomplete fusion of

the parts of the lip.

Inclusions Complete cleft lip – The defect extends through the entire lip

into the floor of the nose.

Incomplete cleft lip – The defect extends through part of the lip

but not into the floor of the nose.

Exclusions Pseudocleft lip – An abnormal linear thickening, depressed

grove, or scar-like pigmentary change on the skin of the lip

without an actual cleft.

Oblique facial clefts

Cleft palate without an associated cleft lip

ICD-9-CM Codes 749.1, 749.2

CDC/BPA Codes 749.10 – 749.29

Diagnostic Methods Cleft lip is usually easily recognized on physical examination

after delivery. It may also be seen on CT or MRI scan, at

surgery or autopsy.

Prenatal Diagnoses Not Confirmed Postnatally While this condition may be identified by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of cleft lip on prenatal ultrasound does not necessarily mean that it will not be

diagnosed after delivery.

Additional Information:

Cleft lip may be unilateral, bilateral, or central in location. If the defect coding system includes unique codes for these different types, the location of the cleft should be coded.

Cleft lip may also be seen in association with amniotic bands. In this instance, the amniotic bands should also be coded.

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Choanal	Atracia
Ciivanai	Autona

Congenital obstruction of the opening of the nasal cavity into **Description**

the nasopharynx on either side. This prevents communication

of the nasal cavity with the pharynx.

Inclusions Choanal atresia, type not specified

Choanal stenosis

Membranous choanal atresia, with or without a bony rim

Completely bony choanal atresia

Exclusions NA

ICD-9-CM Codes 748.0

748.00 **CDC/BPA Codes**

Diagnostic Methods Bilateral choanal atresia is usually easily recognized at birth

> from the clinical presentation of obligate mouth-breathing. Unilateral choanal atresia may be suspected by clinical

examination. Both conditions may be diagnosed by the inability to pass a feeding tube from the nasal passage(s) into the

posterior pharynx. Both conditions may also be seen on CT or

MRI scan, at surgery or autopsy.

Prenatal Diagnoses Not

While these conditions may be identified by prenatal **Confirmed Postnatally** ultrasound, they should not be included in surveillance data

without postnatal confirmation. In addition, the absence of choanal atresia on prenatal ultrasound does not necessarily

mean that it will not be diagnosed after delivery.

Additional Information:

Choanal atresia or stenosis may be unilateral or bilateral. If the defect coding system includes unique codes for these different types, the location should be coded.

Choanal atresia is one of the defects reported as part of the CHARGE association, which may also include colobomas, heart defects, retarded growth and development, genital hypoplasia, and ear anomalies and/or deafness.

Esophageal Atresia/Tracheoesophageal Fistula

Description Esophageal atresia – A condition in which the esophagus ends

in a blind pouch and fails to connect with the stomach.

Tracheoesophageal fistula – An abnormal communication between the esophagus and the trachea. This is almost always

associated with some form of esophageal atresia.

Inclusions Esophageal atresia alone

Esophageal atresia with tracheoesohpageal (TE) fistula

Esophageal stenosis, stricture, ring, or web

TE fistula

Tracheoesophageal fistula, all types

Exclusions Tracheal atresia

Tracheoesophageal cleft

ICD-9-CM Codes 750.3

CDC/BPA Codes 750.30 – 750.35

Diagnostic MethodsThe diagnosis may be suspected by the clinical presentation of

polyhydramnios, vomiting, or respiratory distress. Esophageal atresia may be diagnosed by x-ray documentation of failure of a

feeding tube to pass from the pharynx into the stomach.

Tracheoesophageal atresia may be conclusively diagnosed only

by CT or MRI scan, surgery, or autopsy.

Prenatal Diagnoses Not Confirmed Postnatally

These conditions may be included when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in

the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally before being

included

Additional Information:

In some instances, TE fistula without esophageal atresia may not be diagnosed until weeks, months, or even a year or more after birth if the communication between the esophagus and stomach remains patent.

TE fistula is one of the defects reported as part of the VATER, or VACTERL, association, which may also include vertebral and cardiac defects, anal atresia, renal defects, and limb anomalies.

Rectal and Large Intestinal Atresia/Stenosis

Complete or partial occlusion of the lumen of one or more **Description**

segments of the large intestine and/or rectum.

Inclusions Anal atresia or stenosis

Colonic atresia or stenosis

Imperforate anus

Large intestinal atresia or stenosis

Rectal atresia or stenosis

Exclusions Apple peel intestinal atresia

> Duodenal atresia or stenosis Ileal atresia or stenosis Jejunal atresia or stenosis

Small intestinal atresia or stenosis

751.2 **ICD-9-CM Codes**

751.20 - 751.24**CDC/BPA Codes**

Diagnostic Methods Anal atresia (imperforate anus) is usually easily recognized at

> birth by physical examination. While large intestinal and rectal atresia or stenosis may be suspected by the clinical presentation of failure to pass meconium or stool, they may be conclusively diagnosed only through direct imaging of the bowel by x-ray,

barium enema, surgery, or autopsy.

Prenatal Diagnoses Not

While these conditions may be identified by prenatal ultrasound, they should not be included in surveillance data **Confirmed Postnatally**

without postnatal confirmation. In addition, the absence of intestinal, rectal or anal atresia or stenosis on prenatal ultrasound does not necessarily mean that it will not be

diagnosed after delivery.

Additional Information:

These conditions may occur with or without a fistula.

Anal atresia is one of the defects reported as part of the VATER, or VACTERL, association, which may also include vertebral and cardiac defects, TE fistula, renal defects, and limb anomalies.

Pyloric Stenosis		
Description	Hypertrophy (thickening) of the muscles of the pylorus connecting the stomach to the duodenum, resulting in complete or partial obstruction of the passage of food and gastric contents.	
Inclusions	Infantile (congenital) hypertrophic pyloric stenosis Pyloric stenosis	
Exclusions	Pylorospasm (intermittent spasm of the pyloric muscles) without permanent narrowing of the lumen.	
ICD-9-CM Codes	750.5	
CDC/BPA Codes	750.51	
Diagnostic Methods	Many instances of pyloric stenosis may be diagnosed by the clinical presentation and physical examination. However, other cases may be diagnosed conclusively only by abdominal ultrasound or contrast x-ray of the stomach.	
Prenatal Diagnoses Not Confirmed Postnatally	In rare cases, pyloric stenosis may develop prenatally and may be identified on prenatal ultrasound. However, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of pyloric stenosis on prenatal ultrasound does not mean that it will not develop after delivery (see below).	

Additional Information:

Pyloric stenosis most typically presents with intractable vomiting in a 3- to 4-week-old infant. While it may appear late in gestation, it develops more commonly in the first month or two after birth. As such, it may not be a truly congenital defect.

The etiology of pyloric stenosis remains unclear, but is probably multifactorial with both genetic and environmental influences. Pyloric stenosis has been associated with erythromycin use in newborn infants.

Hirschsprung Disease (Congenital Megacolon)

Description Hirschsprung disease – Absence of the parasympathetic

ganglion nerve cells (aganglionosis) of the wall of the colon or

rectum, which may result in congenital megacolon.

Megacolon – Enlargement of the diameter of part or all of the

colon.

Inclusions Aganglionic megacolon

Congenital megacolon

Hirschsprung disease, type not specified Long-segment Hirschsprung disease (Type II) Short-segment Hirschsprung disease (Type I)

Total colon (intestinal) aganglionosis

Exclusions Psychogenic megacolon

ICD-9-CM Codes 751.3

CDC/BPA Codes 751.30 – 751.34

Diagnostic Methods Hirschsprung disease (congenital megacolon) may be suspected

by contrast x-ray (barium enema). However, it may be diagnosed conclusively only through direct assessment of the

presence or absence of ganglion cells in rectal tissue at biopsy,

surgery, or autopsy.

Prenatal Diagnoses Not Confirmed Postnatally While this condition may be suspected by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of congenital megacolon on prenatal ultrasound does not necessarily mean that it will not

be diagnosed after delivery.

Additional Information:

Megacolon may result from any condition that inhibits normal passage of the intestinal contents. Primary underlying conditions include Hirschsprung disease, rectal and large intestinal atresia/stenosis, imperforate anus, and voluntary stool retention (psychogenic megacolon). In Hirschsprung disease, the aganglionic segment of intestine is small and empty, while the normally enervated segment proximal to the affected area is enlarged and filled with fecal matter.

Hirschsprung disease is classified according to the extent of aganglionosis. In 80% of cases, aganglionosis extends from the anal sphincter and rectum to the middle of the sigmoid colon; in 10% to 20% of cases, it extends further to the transverse or right colon; in 3% of cases, aganglionosis involves the entire colon.

Possible complications of Hirschsprung disease/congenital megacolon include bowel perforation, enterocolitis (intestinal inflammation), peritonitis (inflammation of the lining of the abdomen), and septicemia (bloodstream infection).

Approximately 3% of infants with Down syndrome have aganglionosis of the colon. When Down syndrome and Hirschsprung disease/congenital megacolon occur in the same infant, both conditions should be coded.

Biliary Atresia	
Description	Congenital absence of the lumen of the extrahepatic bile ducts.
Description	<i>8</i>
Inclusions	Agenesis, absence, hypoplasia, obstruction or stricture of the bile duct(s)
Exclusions	Congenital or neonatal hepatitis Intrahepatic biliary atresia (absence or paucity of bile ducts within the liver) not associated with extrahepatic biliary atresia
ICD-9-CM Codes	751.61
CDC/BPA Codes	751.65
Diagnostic Methods	Biliary atresia may be suspected by the clinical presentation and the presence of elevated direct bilirubin and liver function tests. However, it may be conclusively diagnosed only through direct assessment of the bile ducts by abdominal ultrasound, CT or MRI scan, biliary excretion study (HIDA scan), surgery, or autopsy.
Prenatal Diagnoses Not Confirmed Postnatally	While biliary atresia may be suspected by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of biliary atresia on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.

Additional Information:

The liver contains within its substance intrahepatic bile ducts and passages that join and coalesce to form two main ducts that carry bile out of the liver.

The extrahepatic bile ducts include the hepatic duct (formed by the two main ducts that carry bile out of the liver), the cystic duct (which carries bile out of the gallbladder where it is stored), and the common bile duct (formed by the junction of the hepatic duct and the cystic duct), which carries bile into the duodenum for excretion.

When extrahepatic biliary atresia is present, the intrahepatic bile ducts may also be abnormal or atretic.

Patients with biliary atresia may have jaundice due to direct hyperbilirubinemia, which is not treated with phototherapy. The more common type of neonatal jaundice due to indirect hyperbilirubinemia may be treated with phototherapy and does not indicate the presence of biliary atresia.

Renal Agenesis/Hypoplasia

Description Renal agenesis – Complete absence of the kidney

Renal hypoplasia – Incomplete development of the kidney

Inclusions Renal agenesis, dysgenesis, aplasia, or hypoplasia

Potter syndrome secondary to renal agenesis/hypoplasia

Exclusions Cystic renal dysplasia

Cystic kidney disease Multicystic kidney

Multicystic dysplastic kidney

Polycystic kidney Renal cysts Renal dysplasia Small kidney

ICD-9-CM Codes 753.0

CDC/BPA Codes 753.00 – 753.01

Diagnostic MethodsBilateral renal agenesis is usually easily recognized on physical

examination after delivery. Bilateral renal hypoplasia may or may not be recognized after delivery, depending on the severity

and degree of residual kidney function.

Unilateral renal agenesis or hypoplasia may not be symptomatic

at delivery if the contralateral kidney is not impaired.

Each of these diagnoses may be conclusively diagnosed only through direct assessment by abdominal ultrasound, CT or MRI

scan, surgery, or autopsy.

Prenatal Diagnoses Not Confirmed Postnatally Bilateral renal agenesis may be included when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should

always have confirmation of the defect postnatally before being

included.

While bilateral renal hypoplasia and unilateral renal agenesis/hypoplasia may be suspected by prenatal ultrasound, they should not be included in surveillance data without postnatal confirmation. Lack of visualization of a kidney on prenatal ultrasound does not always indicate that the kidney is truly absent.

Additional Information:

Renal agenesis and hypoplasia may be unilateral or bilateral. If the defect coding system includes unique codes for these different types, the location should be coded.

Bilateral renal agenesis, or any condition that significantly impairs the function of both kidneys *in utero*, may lead to the oligohydramnios sequence (Potter syndrome) due to lack of fetal urine production and the resulting decreased amniotic fluid volume. The sequence includes minor facial dysmorphism (flat face, small chin, large ears), pulmonary hypoplasia, and joint contractures.

Bilateral renal agenesis is incompatible with long-term survival unless a kidney transplant is performed. In contrast, unilateral renal agenesis/hypoplasia may not be diagnosed until weeks, months, or even years after birth if the contralateral kidney function is normal. Some unilateral cases may be diagnosed only as incidental findings during evaluation for other conditions, and some may never be recognized.

Bladder Exstrophy		
Description	A defect in the lower abdominal wall and anterior wall of the bladder through which the lining of the bladder is exposed to the outside.	
Inclusions	Classic bladder exstrophy Ectopia vesicae Epispadias-exstrophy complex Extroversion of the bladder Variants of bladder exstrophy Vesical exstrophy	
Exclusions	Ambiguous genitalia without mention of bladder exstrophy Cloacal exstrophy Isolated epispadias	
ICD-9-CM Codes	753.5	
CDC/BPA Codes	753.50	
Diagnostic Methods	Bladder exstrophy is easily recognized on physical examination at delivery. However, the exact nature of the defect and associated anomalies may only be distinguished by abdominal ultrasound, contrast x-ray studies, CT or MRI scan, surgery, or autopsy.	
Prenatal Diagnoses Not Confirmed Postnatally	These conditions may be included when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data, as it may be difficult to distinguish	

Additional Information:

In the classic form of bladder exstrophy, the entire urinary tract is open anteriorly from the urethral meatus to the umbilicus. The pubic bones are widely separated, as are the abdominal muscles and fascia. There is eversion/exposure of the posterior bladder wall. The genitalia of either gender may be involved and may be bifid or duplicated. The classic form of bladder exstrophy occurs more frequently in males.

postnatally before being included.

bladder exstrophy from cloacal exstrophy. Live-born children who survive should always have confirmation of the defect

Variants of bladder exstrophy occur more rarely and affect females more often then males. Included among these variants are superior vesical fistula, closed exstrophy, duplicate exstrophy, pseudoexstrophy, inferior vesicle. Epispadias is almost uniformly present, but should not be coded separately.

Ambiguous genitalia may be noted in patients with bladder exstrophy if an obvious scrotum and

testes are not present. However, ambiguous genitalia should not be coded as a separate defect in these instances.

Bladder exstrophy should be distinguished from cloacal exstrophy, in which the urinary, intestinal, and genital structures open into a common cavity (the cloaca). The distinction may only be possible with detailed diagnostic studies, surgery, or at autopsy. In cloacal exstrophy, bladder exstrophy and imperforate anus are also present. In bladder exstrophy without cloacal exstrophy, the anus is patent. When both bladder and cloacal exstrophy are present, only cloacal exstrophy should be coded.

Appendix 3.2 A3.2-43 Case Definition

Obstructive Genitourinary Defect

Description Partial or complete obstruction of the flow of urine at any level

of the genitourinary tract from the kidney to the urethra.

Inclusions Atresia, stenosis, stricture or occlusion of one or both ureters,

the bladder neck, the urethra or urethral meatus

Dilatation of one or both ureters

Hydronephrosis Hydroureter Hypoplastic ureter Megaloureter

Posterior urethral valves

Obstruction of the ureteropelvic junction (UPJ), the ureterovesical (UV) junction, or the vesicourethral (VU)

junction

Urethral valves, type not specified

Exclusions Inhibition of urinary flow at any of the above sites resulting

solely from neurologic impairment.

ICD-9-CM Codes 753.2, 753.6

CDC/BPA Codes 753.20 – 753.29, 753.60 – 753.69

Diagnostic Methods Genitourinary tract obstruction may be suspected by the clinical

presentation. However, the exact nature of the defect and the level of obstruction may only be distinguished by direct visualization. The upper urinary tract (kidneys and ureters) is usually visualized with renal ultrasound, radionuclide scan, or a contrast study such as intravenous pyelography (IVP). The lower urinary tract (bladder and urethra) is usually visualized directly with cystoscopy or urethral endoscopy, or with contrast

studies such as voiding cystourethrogram (VCUG) and sometimes IVP. Obstructions also may be diagnosed at surgery

or autopsy.

Prenatal Diagnoses Not Confirmed Postnatally While obstructive genitourinary defects may be identified by

prenatal ultrasound, many lesions diminish or resolve

spontaneously prior to birth. For this reason, they should not be included in surveillance data without postnatal confirmation (see below). In addition, the absence of genitourinary

obstruction on prenatal ultrasound does not necessarily mean that an obstructive defect will not be diagnosed after delivery.

Additional Information:

When urine flow is obstructed, the portion of the genitourinary tract proximal to the affected area may become enlarged and dilated with urine. Mild lesions may produce only partial or intermittent urinary obstruction without permanent damage. More severe lesions may substantially or

completely obstruct urine flow, resulting in permanent damage to proximal structures, and sometimes impaired kidney function, if not relieved by surgery.

Increased use of ultrasound screening has led to the recognition of asymptomatic genitourinary tract obstructions in the fetus and newborn, many of which resolve without treatment and would not otherwise have been diagnosed. Inclusion of these lesions in birth defects surveillance data may inflate the apparent frequency of significant obstructive genitourinary defects. If it is possible to correlate the findings on prenatal and/or newborn ultrasound with the clinical course of symptoms and treatment, this should factor into the decision as to which obstructive lesions to include in the surveillance data.

Appendix 3.2 A3.2-45 Case Definition

Hypospadias and Epispadias

Description Hypospadias – Displacement of the opening of the urethra

(urethral meatus) ventrally and proximally (underneath and closer to the body) in relation to the tip of the glans of the

penis.

Epispadias – Displacement of the opening of the urethra (urethral meatus) dorsally and proximally (on the top and closer to the body) in relation to the tip of the glans of the penis.

Inclusions First-degree hypospadias – The urethral meatus is located on

the glans of the penis. Also called primary, 1°, glandular, or

coronal hypospadias.

Second-degree hypospadias – The urethral meatus is located on the shaft of the penis. Also called secondary, 2°, or penile

hypospadias.

Third-degree hypospadias – The urethral meatus is located at the base of the penis on the scrotum or perineum. Also called tertiary, 3°, scrotal, penoscrotal, or perineal hypospadias.

Hypospadias, degree not specified Hypospadias of any type with chordee

Epispadias

Exclusions Chordee alone without associated hypospadias

Ambiguous genitalia

ICD-9-CM Codes Hypospadias 752.61

Epispadias 752.62

CDC/BPA Codes Hypospadias 752.600 – 752.607, 752.620, 752.605 –

752.607

Epispadias 752.621

Diagnostic MethodsBoth hypospadias and epispadias are usually easily recognized

on physical examination at delivery. They may also be seen on

contrast x-rays of the urinary tract, at surgery or autopsy.

While these conditions may be diagnosed by prenatal

Prenatal Diagnoses Not

Confirmed Postnatally ultrasound, they should not be included in surveillance data without postnatal confirmation. In addition, the absence of hypospadias or epispadias on prenatal ultrasound does not

necessarily mean that they will not be diagnosed after delivery.

Additional Information:

Chordee indicates a ventral (downward) curve of the penis, which may result from cutaneous or

fibrous restriction. It is present in approximately 35% to 50% of cases of hypospadias.

In mild forms of first-degree hypospadias, the foreskin may appear hooded but there may be no overt clinical symptoms.

In contrast, third-degree hypospadias may be described as ambiguous genitalia. In this instance, it is important to search the medical record for detailed information (including chromosome, molecular, and hormone analyses; genetics and endocrinology consultations; surgery or autopsy reports) that may clarify the anatomy and/or indicate whether an underlying genetic condition or endocrinopathy associated with ambiguous genitalia is present. Ambiguous genitalia should not be coded if hypospadias is the only diagnosis. Hypospadias generally should not be coded if a normal female karyotype (46,XX) is reported.

Epispadias is almost uniformly present with bladder exstrophy. In these cases, only the bladder exstrophy should be coded.

Reduction Deformity, Upper Limbs

Description

Complete or partial absence of the upper arm (humerus), lower arm (radius and/or ulna), wrist (carpals), hand (metacarpals), or fingers (phalanges).

Inclusions

Transverse limb reduction – Complete or partial absence of the distal (furthest from the body) structures of the arm in a transverse (cross-wise) plane at the point where the deficiency begins. Structures proximal to the point where the deficiency begins remain essentially intact. Types of transverse limb reductions include:

Acheiria – Absence of a hand Adactyly – Absence of digits (fingers), excluding isolated missing thumb (see below) Aphalangia – Absence of phalanges. Fingers contain 3 phalanges each. The thumb contains 2 phalanges.

Amelia – Complete absence of the upper limb (humers, radius, ulna, wrist, hand and fingers).

Hemimelia, Meromelia – Partial absence of a limb. This may refer to either transverse or longitudinal reductions.

Oligodactyly – Fewer than 5 digits.

Transverse terminal deficiency – Complete absence of the distal structures of the arm with the proximal structures intact. This term usually refers to reduction defects below the elbow.

Congenital amputation, type not specified.

Longitudinal limb reduction – Partial absence of the arm in parallel with the long axis of the arm. These may involve preaxial (on the thumb side), postaxial (on the fifth finger side), or central parts of the arm. Types of longitudinal limb reductions include:

Ectrodactyly
Ectromelia
Isolated missing thumb
Lobster claw hand
Radial aplasia or hypoplasia

Split-hand malformation (split hand/split foot malformation, SHSF) – A central longitudinal limb

reduction in which there is complete or partial absence of one or more of the central rays (second through fourth fingers and their associated metacarpal bones) of the hand.

Ulnar aplasia or hypoplasia

Intercalary limb reduction – Complete or partial absence of the proximal (closest to the body) or middle segments of the arm with all or part of the distal segment present.

Phocomelia is a general term for any type of intercalary limb reduction.

Reduction deformities of the upper limb not elsewhere coded or of unspecified type – Complete or partial absence of the arm that does not fall within the above categories or for which there is no specific description.

Exclusions Shortened arms, forearms, hands, or fingers that have all of

their component parts, including those that are part of a generalized chondodystrophy, osteodystrophy, or dwarfism.

Hypoplastic nails

ICD-9-CM Codes 755.20 – 755.29

CDC/BPA Codes 755.20 – 755.29

Diagnostic Methods Limb reductions are usually easily recognized on physical

examination at delivery. However, the exact nature of the defect may only be distinguished by x-ray, surgery, or autopsy.

Prenatal Diagnoses Not

Confirmed Postnatally

While these conditions may be identified by prenatal ultrasound, they generally should not be included in

surveillance data without postnatal confirmation. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Lack of visualization of a bone or limb on prenatal ultrasound does not necessarily mean that the bone or limb truly is not present.

Live-born children who survive should always have

confirmation of the defect postnatally before being included.

Additional Information:

The terminology for limb reduction deformities is often confusing. Some terms (such as "phocomelia"), have been misused and others (such as "ectrodactyly"), have been used for both longitudinal and transverse defects. If medical record review is available, it is important to look for a complete description of all structures that are present and absent in order to verify the diagnosis.

Preaxial refers to the side of the arm on which the thumb and radius are located. Postaxial refers to the side of the arm on which the fifth finger and ulna are located.

Transverse limb reductions may be seen in association with amniotic bands. When both are present, both conditions should be coded.

Rudimentary or nubbin fingers may be present at the distal end of a transverse limb reduction. Their presence alone does not change the classification of the defect as transverse.

Joint contractures are commonly seen in association with longitudinal limb deficiencies.

Intercalary reduction deformities (phocomelia) have been associated with the use of thalidomide during early pregnancy. However, thalidomide use may result in a number of other defects, including longitudinal reduction deformities. Intercalary defects also may occur without exposure to thalidomide.

Reduction deformities are one of the defects that may be reported as part of:

The VATER or VACTERL association, which also may include vertebral, cardiac and renal defects, TE fistula, and anal atresia.

Poland anomaly, which also includes deficiency of the pectoralis muscle on the same side.

Moebius anomaly (Oromandibular-Limb Hypogenesis Spectrum), which also may include a small mouth, small chin (micrognathia), small tongue (hypoglossia), sixth and seventh cranial nerve palsies.

Reduction Deformity, Lower Limbs

Description

Complete or partial absence of the upper leg (femur), lower leg (tibia and/or fibula), ankle (tarsals), foot (metatarsals), or toes (phalanges).

Inclusions

Transverse limb reduction – Complete or partial absence of the distal (furthest from the body) structures of the leg in a transverse (cross-wise) plane at the point where the deficiency begins. Structures proximal to the point where the deficiency begins remain essentially intact. Types of transverse limb reductions include:

Adactyly – Absence of digits (toes) Aphalangia – Absence of phalanges. The smaller toes contain 3 phalanges each. The big toe contains 2 phalanges.

Amelia – Complete absence of the lower limb (femur, tibia, fibula, ankle, foot, and toes).

Hemimelia, Meromelia – Partial absence of a limb. This may refer to either transverse or longitudinal reductions.

Oligodactyly – Fewer than 5 digits.

Transverse terminal deficiency – Complete absence of the distal structures of the leg with the proximal structures intact.

Congenital amputation, type not specified

Longitudinal limb reduction – Partial absence of the leg in parallel with the long axis of the leg. These may involve preaxial (on the big toe side), postaxial (on the fifth toe side), or central parts of the leg. Types of longitudinal limb reductions include:

Ectrodactyly Ectromelia Fibular aplasia or hypoplasia

Split-foot malformation (split hand/split foot malformation, SHSF) – A central longitudinal limb reduction in which there is complete or partial absence of one or more of the central rays (second through fourth toes and their associated metatarsal bones) of the foot.

Tibial aplasia or hypoplasia

Intercalary limb reduction – Complete or partial absence of the proximal (closest to the body) or middle segments of the leg with all or part of the distal segment present.

Phocomelia – A general term for any type of intercalary limb reduction.

Reduction deformities of the lower limb not elsewhere coded or of unspecified type – Complete or partial absence of the leg that does not fall within the above categories or for which there is no specific description.

Exclusions Shortened upper and/or lower legs, feet, or toes that have all of

their component parts, including those that are part of a generalized chondodystrophy, osteodystrophy, or dwarfism.

Hypoplastic nails.

ICD-9-CM Codes 755.30 – 755.39

CDC/BPA Codes 755.30 – 755.39

Diagnostic MethodsLimb reductions are usually easily recognized on physical

examination at delivery. However, the exact nature of the defect may only be distinguished by x-ray, surgery, or autopsy.

Prenatal Diagnoses Not Confirmed Postnatally While these conditions may be identified by prenatal ultrasound, they generally should not be included in surveillance data without postnatal confirmation. However, if it

surveillance data without postnatal confirmation. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Lack of visualization of a bone or limb on prenatal ultrasound does not necessarily mean that the bone or limb truly is not present.

Live-born children who survive should always have

confirmation of the defect postnatally before being included.

Additional Information:

The terminology for limb reduction deformities is often confusing. Some terms (such as "phocomelia") have been misused and others (such as "ectrodactyly") have been used for both longitudinal and transverse defects. If medical record review is available, it is important to look for a complete description of all structures that are present and absent in order to verify the diagnosis.

Preaxial refers to the side of the leg on which the big toe and tibia are located. Postaxial refers to the side of the leg on which the fifth toe and fibula are located.

Transverse limb reductions may be seen in association with amniotic bands. When both are

present, both conditions should be coded.

Rudimentary or nubbin toes may be present at the distal end of a transverse limb reduction. Their presence alone does not change the classification of the defect as transverse.

Joint contractures are commonly seen in association with longitudinal limb deficiencies.

Intercalary reduction deformities (phocomelia) have been associated with the use of thalidomide during early pregnancy. However, thalidomide use may result in a number of other defects, including longitudinal reduction deformities. Intercalary defects also may occur without exposure to thalidomide

Reduction deformities are one of the defects that may be reported as part of:

The VATER or VACTERL association, which also may include vertebral, cardiac and renal defects, TE fistula, and anal atresia.

Moebius anomaly (Oromandibular-Limb Hypogenesis Spectrum), which also may include a small mouth, small chin (micrognathia), small tongue (hypoglossia), sixth and seventh cranial nerve palsies.

Appendix 3.2 A3.2-53 Case Definition

Gastroschisis

A congenital opening or fissure in the anterior abdominal wall **Description**

lateral to the umbilicus through which the small intestine, part of the large intestine, and occasionally the liver and spleen, may herniate. The opening is separated from the umbilicus by a small bridge of skin, and the herniating organs are not covered by a protective membrane. Gastroschisis usually occurs on the right side of the umbilicus, although it may occur on the left.

Inclusions Gastroschisis

Exclusions Omphalocele

ICD-9-CM Codes 756.79

CDC/BPA Codes 756.71

Gastroschisis is usually easily recognized on physical **Diagnostic Methods**

> examination after delivery. However, in some instances, it may be conclusively distinguished from omphalocele only at surgery

or autopsy.

Prenatal Diagnoses Not Confirmed Postnatally

Gastroschisis may be included when only diagnosed prenatally. However, it may be difficult to distinguish gastroschisis from omphalocele on prenatal ultrasound, and the terms sometimes are used interchangeably. If it is possible to ascertain the degree of certainty of the prenatal diagnosis and the location of the umbilical cord insertion relative to the abdominal defect, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally before being included. In addition, the absence of gastroschisis on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.

Additional Information:

The distinction between gastroschisis and omphalocele is important because they have different etiologies and different implications for treatment and long-term survival.

In gastroschisis, the umbilicus and cord are normal and separated from the abdominal wall defect by a small bridge of skin. The herniating organs are not covered by a protective membrane. However, they may appear matted and covered by a thick fibrous material as a result of prolonged exposure to amniotic fluid in utero.

In omphalocele, abdominal organs herniate through the umbilicus into the umbilical cord. There is no bridge of skin between the abdominal wall defect and the umbilicus and cord. While the herniating organs are covered by a protective membrane, this may rupture before, during, or after delivery.

Gastroschisis may be one of the defects reported as part of the Limb-Body Wall complex. This is a disruption complex of the lateral body wall, which may also include limb reductions, neural tube defects, heart defects, and other anomalies.

Maternal serum alphafetoprotein (MSAFP) and/or amniotic fluid alphafetoprotein (AFAFP) may be elevated with gastroschisis. However, these screening tests alone are not sufficient to diagnose the condition.

Omphalocele

Description A defect in the anterior abdominal wall in which the umbilical

ring is widened, allowing herniation of abdominal organs, including the small intestine, part of the large intestine, and occasionally the liver and spleen, into the umbilical cord. The

herniating organs are covered by a nearly transparent

membranous sac.

Inclusions Omphalocele

Exclusions Gastroschisis

Umbilical hernia

ICD-9-CM Codes 756.79

CDC/BPA Codes 756.70

Diagnostic Methods Omphalocele is usually easily recognized on physical

examination after delivery. However, in some instances, it may be conclusively distinguished from gastroschisis only at surgery

or autopsy.

Prenatal Diagnoses Not Confirmed Postnatally Omphalocele may be included when only diagnosed prenatally. However, it may be difficult to distinguish omphalocele from gastroschisis on prenatal ultrasound, and the terms sometimes are used interchangeably. If it is possible to ascertain the degree of certainty of the prenatal diagnosis and the location of the umbilical cord insertion relative to the abdominal defect, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally before being included. In addition, the absence of omphalocele on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.

Additional Information:

The distinction between omphalocele and gastroschisis is important because they have different etiologies and different implications for treatment and long-term survival.

In omphalocele, abdominal organs herniate through the umbilicus into the umbilical cord. There is no bridge of skin between the abdominal wall defect and the umbilicus and cord. While the herniating organs are covered by a protective membrane, this may rupture before, during, or after delivery.

In gastroschisis, the umbilicus and cord are normal and separated from the abdominal wall defect by a small bridge of skin. The herniating organs are not covered by a protective membrane. However, they may appear matted and covered by a thick fibrous material as a result of prolonged exposure to amniotic fluid *in utero*.

Omphalocele is one of the defects reported as part of the Omphalocele-Exstrophy-Imperforate Anus-Spina Bifida (OEIS) complex.

Maternal serum alphafetoprotein (MSAFP) and/or amniotic fluid alphafetoprotein (AFAFP) may be elevated with omphalocele. However, these screening tests alone are not sufficient to diagnose the condition.

In contrast to omphalocele, umbilical hernias are completely covered by normal skin.

Conge	nita	lΗ	lip	Dis	loc	atio	n

Description Location of the head of the femur (bone of the upper leg)

outside its normal location in the cup-shaped cavity formed by

the hip bones (acetabulum).

Inclusions Congenital hip dislocation, unilateral or bilateral

Developmental dysplasia of the hip

Teratologic hip dislocation

Exclusions Flexion deformity/contracture of the hip

Hip click

Predislocation of the hip Preluxation of the hip Subluxation of the hip

Unstable hip

ICD-9-CM Codes 754.30, 754.31, 754.35

CDC/BPA Codes 754.30

Diagnostic Methods Hip dislocation may be suspected, and sometimes diagnosed,

by physical examination. However, ultrasound or x-ray are the

definitive diagnostic tests.

Prenatal Diagnoses Not Confirmed Postnatally While this condition may be suspected by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of hip dislocation on prenatal ultrasound does not necessarily mean that it will not be

diagnosed after delivery.

Additional Information:

The terminology describing congenital hip dislocation is often confusing. An unstable hip, in which the femoral head may be moved in and out of the acetabulum on physical examination, often resolves spontaneously in young infants. A truly dislocated hip, in which the femoral head remains out of the acetabulum for a prolonged period, may result in acetabular deformity unless treated. Hence, the designation developmental dysplasia of the hip.

The stability of the hip joint may be evaluated on physical examination. In the Barlow test, lateral pressure is applied to the hip with the knees flexed in an attempt to move the head of the femur out of the hip joint (acetabulum) into a dislocated position. In the Ortolani maneuver, a laterally dislocated femoral head is moved back into normal position in the acetabulum by applying pressure medially. The presence of either sign indicates a hip dislocation is present. However, their absence does not always mean that a dislocation is not present. In some instances, the femoral head may be fixed in a dislocated position and it may not be possible to move it in and out of the joint.

Congenital hip dislocation occurs more frequently after footling or breech deliveries and is more common in females than males. It is most often an isolated condition, although hip dysplasia may occur with generalized skeletal abnormalities and in some genetic syndromes. Some instances of congenital hip dislocation are probably familial.

	Diaphragmatic Hernia
Description	Incomplete formation of the diaphragm through which a portion of the abdominal contents herniate into the thoracic cavity.
Inclusions	Absence of the diaphragm Bochdalek hernia – Herniation through a defect in the posterolateral portion of the diaphragm.
	Diaphragmatic hernia, type not specified Hemidiaphragm
	Morgagni hernia – Herniation through a defect in the anterior portion of the diaphragm.
	Paraesophageal hernia – Herniation through a defect in the central portion of the diaphragm surrounding the esophagus.
Exclusions	Eventration of the diaphragm – Weakness in, or absence of, the muscles of the diaphragm which allows upward displacement of a portion of the abdominal contents. However, there is no true herniation of contents through the diaphragm into the thoracic cavity.
ICD-9-CM Codes	756.6
CDC/BPA Codes	756.610 – 756.617
Diagnostic Methods	While diaphragmatic hernia may be suspected by the clinical presentation of respiratory distress, feeding intolerance, and/or cardiac compromise, it may be conclusively diagnosed only through x-ray, contrast study of the bowel, CT or MRI scan, surgery, or autopsy.
Prenatal Diagnoses Not Confirmed Postnatally	Diaphragmatic hernia may be included in surveillance data when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally before being included.

Additional Information:

Children with diaphragmatic hernia often have accompanying abnormalities of the heart, intestine, and lungs, including hypoplastic lungs, which result from the abnormal location of abdominal organs within the thoracic cavity during development.

Trisomy 13

Description The presence of three copies of all or a large part of

chromosome 13.

Inclusions Patau syndrome

Mosaic Patau syndrome Mosaic trisomy 13

Translocation Patau syndrome Translocation trisomy 13

Trisomy 13, not otherwise specified Trisomy D₁, not otherwise specified

Exclusions Balanced translocations involving chromosome 13

ICD-9-CM Codes 758.1

CDC/BPA Codes 758.10 – 758.19

Diagnostic Methods Trisomy 13 may be suspected on physical examination.

However, it may be diagnosed conclusively only through direct

analysis of the infant's chromosomes (karyotype). The chromosomes may be obtained from blood or tissue cells.

Prenatal Diagnoses Not Confirmed Postnatally Trisomy 13 may be included when only diagnosed through direct analysis of fetal chromsomes obtained from amniocentesis, chorionic villus sampling (CVS), or percutaneous umbilical blood sampling (PUBS). However, when mosaic trisomy 13 is noted, the defect should be confirmed postnatally on a specimen obtained directly from the

infant or fetus after birth (see below).

Additional Information:

When the two copies of chromosome 13 from one parent do not separate during egg or sperm formation, three copies of the entire chromosome 13 will be present in the fetus. In this instance, the karyotype is written as 47,XX,+13 or 47,XY,+13. This is the most common type of trisomy 13 and is associated with advanced maternal age, particularly of 35 years or greater.

Translocation trisomy 13 occurs when two separate copies of chromosome 13 are present, but a third copy of part of chromosome 13 is attached to another chromosome. In this instance, there are 46 total chromosomes present, but 3 copies of part of chromosome 13.

Mosaic trisomy 13 occurs when some, but not all, of the cells in the body contain three copies of all or a large part of chromosome 13. In this instance, the karyotype is written as 46,XY/47,XY,+13, for example. Because the placenta may contain mosaic cell lines not present in the fetus, mosaic trisomy 13 diagnosed through chorionic villus sampling should always be confirmed by direct examination of fetal chromosomes from amniocentesis, PUBS, or preferably postnatal blood or tissue samples.

Approximately 80% of infants with trisomy 13 do not survive beyond the first month of life. Major malformations associated with trisomy 13 may include holoprosencephaly, microcephaly, meningomyelocele, cleft lip and/or palate, microphthalmia, retinal dysplasia, polydactyly, heart defects (most commonly a VSD), omphalocele, and genitourinary defects, among others. Among children who survive the newborn period, severe developmental delay is virtually always present as may be deafness, visual impairment, minor motor seizures, and apneic spells.

Infants with mosaic trisomy 13 may be less severely affected with variable degrees of developmental delay and longer survival. Infants with partial trisomy for the proximal segment of chromosome 13 (13pter \rightarrow q14) exhibit a nonspecific pattern of abnormalities with near-normal survival. Approximately 25% of infants with partial trisomy for the distal segment of chromosome 13 (13q14 \rightarrow qter) die during early postnatal life.

Children who survive exhibit severe developmental delay and specific abnormalities.

Major malformations that occur with trisomy 13 in the same infant should be coded separately, as their presence may varies among affected individuals.

Appendix 3.2 A3.2-61 Case Definition

Down Syndrome (Trisomy 21)

Description The presence of three copies of all or a large part of

chromosome 21.

Inclusions Down syndrome

Mosaic Down syndrome Mosaic trisomy 21

Translocation Down syndrome Translocation trisomy 21

Trisomy 21, not otherwise specified

Exclusions Balanced translocations involving chromosome 21

"Downs facies" without associated trisomy 21.

ICD-9-CM Codes 758.0

CDC/BPA Codes 758.00 – 758.09

Diagnostic Methods Down syndrome may be suspected on physical examination.

However, it may be diagnosed conclusively only through direct

analysis of the infant's chromosomes (karyotype). The chromosomes may be obtained from blood or tissue cells.

Prenatal Diagnoses Not Confirmed Postnatally Down syndrome may be included when only diagnosed through direct analysis of fetal chromsomes obtained from amniocentesis, chorionic villus sampling (CVS), or percutaneous umbilical blood sampling (PUBS). However, when mosaic trisomy 21 is noted, the defect should be confirmed postnatally on a specimen obtained directly from the

infant or fetus after birth (see below).

Additional Information:

When the two copies of chromosome 21 from one parent do not separate during egg or sperm formation, three copies of the entire chromosome 21 will be present in the fetus. In this instance, the karyotype is written as 47,XX,+21 or 47,XY,+21. This is the most common type of trisomy 21 and is associated with advanced maternal age, particularly of 35 years or greater.

Translocation trisomy 21 occurs when two separate copies of chromosome 21 are present, but a third copy of part of chromosome 21 is attached to another chromosome. In this instance, there are 46 total chromosomes present, but 3 copies of part of chromosome 21.

Mosaic trisomy 21 occurs when some, but not all, of the cells in the body contain three copies of all or a large part of chromosome 21. In this instance, the karyotype is written as 46,XY/47,XY,+21, for example. Because the placenta may contain mosaic cell lines not present in the fetus, mosaic trisomy 21 diagnosed through chorionic villus sampling should always be confirmed by direct examination of fetal chromosomes from amniocentesis, PUBS, or preferably postnatal blood or tissue samples.

Infants with Down syndrome have a typical appearance and other characteristics, including decreased muscle tone (hypotonia), a weak startle (Moro) reflex, hyperflexible joints, a flattened facial profile, upslanting eyes, abnormally shaped external ears (auricles), loose skin on the back of the neck, dysplasia of the pelvic bones, incurving of the fifth finger (clinodactyly), and a single transverse crease in the palm of the hand (Simian crease). Developmental delay is virtually always present. Major malformations associated with Down syndrome include heart defects (most notably endocardial cushion defects), gastrointestinal defects, and vertebral abnormalities, among others.

Major malformations that occur with Down syndrome in the same infant should be coded separately, as their presence may varies among affected individuals.

Mongolism is an outdated term for Down syndrome.

Trisomy 18

Description The presence of three copies of all or a large part of

chromosome 18.

Inclusions Edwards syndrome

Mosaic Edwards syndrome

Mosaic trisomy 18

Translocation Edwards syndrome

Translocation trisomy 18

Trisomy 18, not otherwise specified

Exclusions Balanced translocations involving chromosome 18

ICD-9-CM Codes 758.2

CDC/BPA Codes 758.20 – 758.290

Diagnostic MethodsTrisomy 18 may be suspected on physical examination.

However, it may be diagnosed conclusively only through direct

analysis of the infant's chromosomes (karyotype). The chromosomes may be obtained from blood or tissue cells.

Prenatal Diagnoses Not Confirmed Postnatally Trisomy 18 may be included when only diagnosed through direct analysis of fetal chromsomes obtained from amniocentesis, chorionic villus sampling (CVS), or percutaneous umbilical blood sampling (PUBS). However, when mosaic trisomy 13 is noted, the defect should be confirmed postnatally on a specimen obtained directly from the

infant or fetus after birth (see below).

Additional Information:

When the two copies of chromosome 18 from one parent do not separate during egg or sperm formation, three copies of the entire chromosome 18 will be present in the fetus. In this instance, the karyotype is written as 47,XX,+18 or 47,XY,+18. This is the most common type of trisomy 18 and is associated with advanced maternal age, particularly of 35 years or greater.

Translocation trisomy 18 occurs when two separate copies of chromosome 18 are present, but a third copy of part of chromosome 18 is attached to another chromosome. In this instance, there are 46 total chromosomes present, but 3 copies of part of chromosome 18.

Mosaic trisomy 18 occurs when some, but not all, of the cells in the body contain three copies of all or a large part of chromosome 18. In this instance, the karyotype is written as 46,XY/47,XY,+18, for example. Because the placenta may contain mosaic cell lines not present in the fetus, mosaic trisomy 18 diagnosed through chorionic villus sampling should always be confirmed by direct examination of fetal chromosomes from amniocentesis, PUBS, or preferably postnatal blood or tissue samples.

Most pregnancies affected with trisomy 18 result in spontaneous abortion. Approximately 50% of

live-born infants with trisomy 18 do not survive beyond the first week of life. Only 5% to 10% survive beyond the first year of life. Major malformations associated with trisomy 18 may include microcephaly, micrognathia, cleft lip and/or palate, heart defects, omphalocele, and renal defects, among others. Minor anomalies associated with trisomy 18 may include low-set malformed auricles (external ears), overlapping of the index and fifth fingers over the third and fourth fingers, absent distal crease on the fifth finger, hirsutism (excess hair) of the forehead and back, lateral deviation of the hands, a hypoplastic thumb, a single transverse palmar crease, and rocker-bottom feet, among others. Developmental delay is virtually always present, as may be hypertonicity, a weak cry, growth retardation, hypoplasia of skeletal muscle and subcutaneous fat, and clenched hands.

Infants with mosaic trisomy 18 may be less severely affected, with variable degrees of developmental delay and longer survival. Infants with trisomy of only the short arm of chromosome 18 (partial trisomy 18) exhibit a nonspecific pattern of abnormalities with mild to no developmental delay. Infants with trisomy of the short arm, centromere, and proximal third of the long arm of chromosome 18 exhibit features of trisomy 18 but not the entire spectrum of abnormalities. Infants with trisomy of only one-third to one-half of the long arm of chromosome 18 exhibit features of trisomy 18 but have longer survival and less severe developmental delays.

Major malformations that occur with trisomy 18 in the same infant should be coded separately, as their presence varies among affected individuals.

Fetal Aclohol Syndrome (FAS)

Description A spectrum of abnormalities resulting from exposure to alcohol

in utero. While the specific abnormalities vary among individuals, the hallmarks include growth deficiency,

microcephaly, facial dysmorphisms, and neurodevelopmental

abnormalities.

Inclusions Fetal alcohol syndrome (FAS)

Exclusions Fetal alcohol effects/facies, without diagnosis of FAS

ICD-9-CM Codes 760.71

CDC/BPA Codes 760.71

Diagnostic Methods While fetal alcohol syndrome may be suspected from a history

of maternal alcohol use during pregnancy, the condition may be conclusively diagnosed only through direct examination of the

infant by a physician (usually a dysmorphologist or

developmental specialist) familiar with the spectrum of FAS

abnormalities.

Prenatal Diagnoses Not Confirmed Postnatally While fetal alcohol syndrome may be suspected from a history of maternal alcohol use during pregnancy, the condition should

not be included in surveillance data without postnatal

confirmation.

Additional Information:

A number of minor malformations may be present with fetal alcohol syndrome, most notably hypoplasia of the maxillary bone (middle) of the face, and a thin upper lip with smooth philtrum (crease). However, these are often subtle in the newborn and may not be recognized until later in childhood. Older children with FAS may manifest poor coordination, irritability, hyperactivity, and neurodevelopmental abnormalities.

Fetal alcohol syndrome is the extreme of a spectrum of effects on growth and development resulting from alcohol exposure *in utero*. At low levels of exposure, the only apparent effect may be a reduction in birth weight. The clinical features and neurodevelopmental abnormalities become increasingly prominent with increasing levels of exposure.

Amniotic Bands

Description Strands of tissue that float in the amniotic fluid as a

consequence of tears or ruptures in the amniotic membrane

which surrounds the fetus during development.

Inclusions Amniotic bands

Amniotic band sequence, syndrome, or disruption complex

Amniotic rupture sequence

Streeter bands

Constriction rings – Soft tissue depressions or grooves

encircling part of the body, usually a limb.

Exclusions NA

ICD-9-CM Codes NA

CDC/BPA Codes 658.80

Diagnostic Methods Structural defects resulting from amniotic bands usually are

readily apparent on physical examination after delivery. However, the fact that they are a consequence of amniotic bands may not be apparent unless a remnant of an amniotic strand is present or amniotic bands were noted on prenatal

ultrasound.

Prenatal Diagnoses Not Confirmed Postnatally

When amniotic bands are seen on prenatal ultrasound, their presence should be correlated with any structural defects noted, and the guidelines for including those defects when only

and the guidelines for including those defects when only diagnosed prenatally should be followed. Live-born children who survive should always be examined for evidence of amniotic bands postnatally. In addition, the absence of amniotic bands on prenatal ultrasound does not necessarily mean that

they are not truly present.

Additional Information:

Amniotic bands may be present in the amniotic sac without impacting the fetus. When noted as an isolated condition without associated structural defects, they should not be coded.

Structural defects that may occur as a result of amniotic bands include:

Pseudosyndactyly (digits compressed together by an encircling band)

Distal limb amputation, hypoplasia, lymphedema, or deformation

Oral clefts

Encephalocele

Anencephaly

Other disruptive defects of the skull

Appendix 3.3

Examples of Conditions Considered to Be Minor Anomalies

Appendix 3.3 Examples of Conditions Considered to Be Minor Anomalies²

Eye

- > Epicanthal folds
- ➤ Iris freckles, Brushfield spots
- > Upward or downward palpebral slant

Ear

- > Darwinian point or tubercle
- > Thickened or excessively folded helix
- ➤ Lack of helical folding
- > Creased, notched, or bifid ear lobe
- ➤ Lop, cup-shaped, or retroverted ear
- > Preauricular sinus, cyst, pit, or skin tag

Head, Face and Neck

- > Flat occiput
- > Frontal bossing
- ➤ Flat brow
- > Flat or prominent bridge of nose
- > Anteverted nostrils
- ➤ Long nasal septum
- > Webbed or redundant neck skin

Hands and Feet

- > Single or horizontal palmar crease
- Clinodactyly
- > Tapered fingers
- Overlapping digits
- ➤ Webbed or widely spaced 2nd and 3rd toes
- > Prominent heel

Other

- Sacral dimples
- ➤ Nevi
- ➤ Cafe-au-lait spots
- Mongolian spots
- Accessory nipples
- > Umbilical hernia
- Vaginal tag
- ➤ Single umbilical artery

Appendix 3.3 A3.3-1 Case Definition

² This is not a comprehensive list. The exact abnormalities considered to be minor defects may vary among experts.

Appendix 3.4

Conditions Related to Prematurity in Infants Born at Less Than 36 Weeks Gestation

Appendix 3.4 Conditions Related to Prematurity in Infants Born at Less Than 36 Weeks Gestation

- Dolichocephaly
- Scaphocephaly
- ➤ Blue sclera
- > Fused eyelids
- ➤ Absent or decreased ear cartilage
- > Patent foramen ovale
- > Patent ductus arteriosus
- ➤ Hypoplastic lungs
- > Small or hypoplastic nipples
- > Hypoplastic labia majora
- > Undescended testicles
- Inguinal hernia

Chapter 4 Data Variables

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4.1 Introduction

The potential data sources available to birth defects programs contain a wide variety of information. Each item of information a birth defects program collects requires staff time to locate, abstract, code, and evaluate, as well as computer space to store it. Thus, due to limited resources, a birth defects program must be efficient in the scope of the information it collects and the manner in which the information is collected and stored.

In this chapter we discuss a number of issues relating to the data variables that comprise a birth defects surveillance system. In Section 4.2, for example, we discuss the criteria that should be considered in selecting the variables that will be collected by a surveillance system. In Section 4.3, we present the three possible origins of surveillance data variables; that is, variables may be abstracted, derived or created. Other topics include possible formats for data variables (Section 4.4), logic checks that can be used to ensure data fall within an expected range (Section 4.5), sources for data variables (Section 4.6), and issues concerning a subset of variables related to birth defects risk factors (Section 4.7). In Section 4.8, we introduce two tables that summarize core (Table 4.1) and recommended (Table 4.2) data variables for a birth defects surveillance system. Additional detail on each of these core and recommended variables is provided in Appendices 4.1 and 4.2, respectively.

It is our hope that the information in this chapter of *The Surveillance Guidelines* will promote and guide standardization of data elements across birth defects surveillance programs. Using standard data elements is particularly important when aggregating data for regional or national analysis. Standardization allows and supports comparisons and collaborations between states.

Whether a surveillance program is based on active or passive case ascertainment, our recommendation is that vital records information or copies (including birth, death or fetal death certificates as appropriate) be obtained. This allows the collection of some data using sources from which population-based demographic information can also be obtained.

Note that we are indebted to Lynberg and Edmonds (1994) for much of the information in this chapter.

Chapter 4 4-1 Data Variables

4.2 Criteria to be Considered in Selection of Data Variables

A birth defects program should consider a number of different criteria when deciding which variables to collect. These include type of case ascertainment, program objectives, and data characteristics. Each of these criteria is discussed further below. The criteria considered in compiling the lists of core and recommended variables are summarized for each variable under the heading 'Justification' in Appendices 4.1 and 4.2.

4.2.1 Type of Case Ascertainment

The case identification methods used by a surveillance program may place constraints on the data variables collected. The available data source(s) for program variables are determined primarily by these methods. For example, birth certificate files usually offer limited data for diagnostic confirmation of the birth defect or a precise description of the defect. An infant's medical record, other than the newborn record, is not likely to include data on the prenatal care received by the mother (see Chapter 6 on Case Ascertainment Methods).

4.2.2 Program Objectives

A surveillance program should limit the information collected to those items needed to fulfill its stated objectives. However, it can be difficult to determine what constitutes this essential information. Often individuals, groups, or organizations that utilize surveillance information may request data on variables that are not really needed and will not be used. One guideline a surveillance program might follow is that information should not be collected if it does not serve at least one programmatic objective.

CDC defines *surveillance* as "the ongoing systematic collection, analysis, and interpretation of health data essential to the planning, implementation and evaluation of public health practice, closely integrated with the timely dissemination of these data to those who need to know" (Centers for Disease Control and Prevention, 1988). Under this definition, it is clear there are a number of functions and objectives for which a birth defects program might need to collect data:

- > Descriptive epidemiology and monitoring. Data can be examined to determine and describe the distribution of a disease (condition) within a population along the parameters of place, person, and time. Monitoring offers quantitative estimates of the magnitude of the disease.
- Research. Data can be used to test hypotheses or in planning research to learn the causes of a disease.
- > Service/planning. Increasingly, surveillance programs are using information on newly identified children with birth defects to refer them for services. These include specialized medical care, educational and early intervention programs, and genetic counseling. Data can also be applied to evaluate services and prevention measures within a population. Knowledge about the disease or condition and changes in the population can assist in optimizing available resources and services.
- Linkage. Variables may be used to link to other databases such that data in those databases may be associated at the case level to complement and enrich case-specific data. Linkage is also an essential surveillance management tool needed to identify and consolidate duplicates.

Chapter 4 4-2 Data Variables

4.2.3 Data Characteristics

Among the important data characteristics a surveillance program should consider are availability, consistency, accuracy, uniqueness, definability, collectability, and comparability. We discuss each of these in turn below.

- Availability. Data must be retrievable from the data sources and be available to the birth defects program. Many data variables are collected and stored at data sources in clinical and administrative databases, facilitating availability and retrievability. In most cases, information should only be collected if it is consistently available. This is particularly true if the information is to be used for statistical analyses or for identifying or contacting case families. If information can be found only in a small portion of the data sources, then staff will spend considerable time looking for unavailable information. The birth defects program may want to either limit collection of such information or work to identify a data source where the same information is consistently available. An exception to this may be where the information is important even if it is only occasionally found in the data sources (e.g., the fact that the infant is in foster care or has been placed for adoption). However, as noted before, this information may be difficult to find and time-consuming to collect.
- Consistency. It is important that the information assembled within the surveillance system has a consistent meaning from report to report. When obtaining information from a range of data sources, it is essential to have a usable level of consistency from source to source. This is especially important for passive data collection and data mining. Simple issues, such as field content and even field size, can significantly affect the comparability and usefulness of the data. Coding rules and practices are special areas of concern.
- Accuracy. The information collected should be accurate. If the information is of questionable veracity, then it should not be collected. Second-hand information found in medical records may be incomplete or inaccurate. If information such as medication use and exposures is important, it should be collected from a reliable source, such as through direct contact with the mother, rather than from medical records.
- > Uniqueness. Programs should avoid the collection of redundant information. Information should not have to be recorded in more than one field. For example, if the infant or fetus delivery date and the mother's date of birth are collected, then the mother's age at delivery does not need to be collected
- Definability. There should be clear definitions for each of the data variables a birth defects program collects.
- ➤ Collectability. The data variables should lend themselves to easy abstraction. This is a potential problem with complex or subjective information. If it takes an excessive amount of time to track down and collect the information, or if there is a high degree of inter-staff variability in how the information is collected, then the information recorded in the birth defects program's database will be of dubious quality and reliability (Horwitz and Yu, 1984; Demlo et al., 1978). In addition, extensive efforts may be necessary for quality control.
- Comparability. The birth defects program may want to consider whether other birth defects programs have access to the same sources and types of data. If the program uses a unique data source or collects a unique data variable that other birth defects programs do not, then the program may not be able to compare its data to those of other programs. This may be of limited importance, however, if the data are being collected to meet specific programmatic objectives, where comparison between different states or programs is unimportant.

Chapter 4 4-3 Data Variables

4.3 The Origins of Data Variables

Data variables may be abstracted, derived, or created.

- ➤ Abstracted data variables. These are data that are available only from the data sources, and the data sources must supply them.
- ➤ Derived data variables. Some data variables are not collected directly from data sources but are rather derived from other information collected from the data sources, e.g., census tract numbers, standardized geographic tables, disease codes.
- > Created data variables. Some data variables may need to be created by the birth defects program, e.g., unique case and staff IDs.

Some data variables may fall into more than one of the above categories. For example, if the mother's age at delivery is not available from the data sources, it may be derived using the date of delivery and the mother's date of birth. The origins of each of the core and recommended variables are summarized under the heading 'Source' in Appendices 4.1 and 4.2.

Chapter 4 4-4 Data Variables

4.4 The Formats of Data Variables

Data may be stored in a computer database in a variety of formats, including as a numerical field, a date field, a text field, a checkbox, or a coded data field. Each of these formats is briefly described below. The format for each of the core and recommended variables is also summarized under the heading 'Type' in Appendices 4.2 and 4.2.

- ➤ Numerical field. A field that includes only numbers.
- ➤ Date field. A field that includes only dates, which are comprised of month, day, and year in a variety of orders and combinations.
- Text field. A field that can contain letters, numbers, and punctuation. Text fields are often of a fixed width. Text fields of infinite width are often called Amemo@fields.
- > Checkbox. A field that contains only two options yes/no, on/off.
- > Coded data field. Data may be collected and stored as they appear in the data source, or they may be 'coded'. A code may contain numbers or letters or both. Whether a birth defects program collects and stores data as coded or not depends on the types of data, as well as on potential uses.

If a birth defects program plans to use a field for analysis, then it is important that the field be easily coded or categorized, permitting ready analysis rather than having to sort through a large collection of free-form text. This is because information such as race/ethnicity, diagnoses, and conditions can be described in a number of different ways. For example, a person may be described as 'African-American' or 'black'. A 'cleft lip' may also be described as a 'lip cleft' or a 'harelip'.

Coding eliminates the problem of having to sort through a variety of differing descriptions. It allows for timely and efficient analysis of data and referral of cases. Coding also enables researchers to know that they are talking about the same thing, and it allows for comparability between different birth defects programs using the same or comparable coding systems.

Whenever possible, a birth defects program should use coding systems consistent or compatible with those used by other groups, particularly other birth defects programs, thus allowing for efficient comparison of data. This applies not only to diagnostic codes but also to characteristics such as maternal race and ethnicity.

Chapter 4 4-5 Data Variables

4.5 Data Variable Logic Checks

Errors may occur in the data collection by a birth defects program, either because of errors in data listed in the data source or because of errors in abstraction. A birth defects program should have some method to identify and correct errors (see Chapter 7 on Data Quality Management). One means of identifying and correcting errors is through *logic checks* that ensure data occur within expected ranges.

Many of the core variables in a birth defects surveillance system have a limited number of options or ranges of values. For example, a gestational age of 75 weeks is highly unlikely to occur. And other variables may have certain logical relationships to one another. For example, the mother's date of birth must always be earlier than the infant's date of birth.

Suggested logic checks for each of the core and recommended variables are summarized under the heading 'Checks' in Appendices 4.1 and 4.2.

Chapter 4 4-6 Data Variables

4.6 Data Variable Location

A birth defects surveillance program may have access to a variety of data sources and will collect data on a number of different variables. Clearly, the same variable may be available from several sources. Abstracting data from a variety of sources allows for greater thoroughness in data collection. If a variable is missing in one data source, it may be available in another source.

Staff collecting data should know where a given data variable is likely to be found, as well as the prioritization of sources for those variables retrievable from multiple data sources, since data sources may disagree as to the value for a particular variable. For example, the infant's delivery medical record and the birth certificate might record different values for birth weight. A birth defects program should prioritize the data sources for particular variables. In the above instance, for example, a birth defects program may decide that the birth weight in the medical record takes precedence over the birth weight from a birth certificate.

For each of the core and recommended variables, the data source – as well as the location within the data source where the variable is most likely to be consistently found – are summarized under the heading 'Location' in Appendices 4.1 and 4.2.

Chapter 4 4-7 Data Variables

4.7 Risk Factor Variables

Risk factors in birth defects include: conditions, illnesses, or complications during pregnancy, labor, or delivery

Selected conditions, such as maternal diabetes and thyroid disease, have been associated with increased risk for certain birth defects (Becerra et al., 1990; Khoury et al., 1989). Information on conditions and complications during pregnancy and delivery may be useful for making syndromic classifications or identifying causality of birth defects, such as diabetic embryopathy.

However, there are a large number of conditions and complications possible during pregnancy and delivery, and birth defects programs could create lists of dozens to hundreds of them. Such long lists would require additional computer storage space and training of field staff regarding where to find the information and how to collect it. Even then, confusion may ensue over which conditions and complications to abstract and subjective differences between staff in their abstraction of this information. Moreover, the information in the data sources commonly available to birth defects programs may not necessarily be consistent or accurate (Olson et al., 1997).

For all of these reasons, birth defects surveillance programs should give careful consideration to the potential thoroughness and usefulness of routine data collection regarding risk factors as relevant to their goals and objectives. In general, programs are more likely to obtain useful information on conditions and complications during pregnancy and delivery through contact with parents, as is done in case-control research studies, than through medical records abstraction.

Chapter 4 4-8 Data Variables

4.8 Data Variable Tables

In the late 1980s, before creation of the National Birth Defects Prevention Network, Larry Edmonds of the Centers for Disease Control and Prevention (CDC) – along with F. John Meaney of Arizona and Susan Panny of Maryland and others – collaborated on development of a set of core data items relevant to birth defects surveillance (Edmonds et al., 1988), based on an earlier list developed by CDC's National Center for Health Statistics. We used the list developed by Edmonds et al. as the foundation for developing the current list of data variables that the NBDPN recommends for birth defects surveillance programs, adding a number of different variables in order to reflect the fact that birth defects surveillance programs have evolved considerably since the 1980s into programs with a variety of objectives and multiple areas of interest.

The data variables in Tables 4.1 and 4.2 (as well as in their corresponding appendices) are categorized as to whether they are infant, maternal, paternal, or contact information variables. For each data variable, we also note in Tables 4.1 and 4.2 the usefulness of that data item relative to a program's specific objectives, which may include descriptive epidemiology and monitoring, research, service and planning, and linkage capability (see Section 4.2.2. for further discussion of program objectives).

To provide a sense of the relative importance of the data variables for a new or expanding surveillance program, we have further distinguished between minimum (or core) variables (Table 4.1 and Appendix 4.1) and recommended variables (Table 4.2 and Appendix 4.2).

- ➤ *Minimum (core) variables* are those that are considered necessary to fulfill the most basic programmatic objectives and that also meet most or all of the supplemental criteria discussed earlier in this chapter.
- **Recommended variables** are those that have the potential to enhance surveillance capability or to support broader programmatic objectives.

By glancing down the column for a specific programmatic objective (e.g., 'research'), the reader can determine – based on the relevant check marks – which elements are considered 'core' and which other data elements are 'recommended' to support a given program objective. These data variables can be abstracted using a minimum number of data sources, including maternal records, infant records, and vital records. Birth defects programs that use the passive case ascertainment approach will find the vital record particularly useful as a data source for many of the maternal core data variables.

After reviewing these lists, birth defects surveillance staff may also wish to add further data variables they consider essential for their own specific programmatic purposes.

Chapter 4 4-9 Data Variables

Table 4.1 Minimum (Core) Data Variables

Data Variable	Descriptive Epidemiology and Monitoring	Research	Service/ Planning	Linkage	
	Infan				
Unique ID	✓	✓	✓	✓	
Date of Pregnancy Outcome	✓	✓	✓	✓	
Sex	✓	✓	✓	✓	
Infant's Name					
First	✓.	✓.	✓.	✓.	
Middle	✓.	√	√	√	
Last	√	√	√	√	
Suffix	✓	✓	✓	✓	
Source of Report	✓	√	✓	✓	
Medical Record Number(s)	✓	✓	✓	✓	
Vital Record Certificate Number				✓	
Place of Pregnancy Outcome	✓	✓	✓	✓	
Pregnancy Outcome	✓	✓	✓	✓	
Birth Weight	✓	✓	✓	✓	
Plurality	✓	✓	✓	✓	
Gestational Age	✓	✓	✓	✓	
Diagnosis Code	✓	✓	<	<	
	Contact Info	rmation			
Name of Responsible Party			✓		
Address of Responsible Party			√		
Telephone Number of Responsible Party			√		
Mother Mother					
Mother's Date of Birth	✓	✓	✓	✓	
Mother's Race	✓	✓			
Mother's Ethnicity	✓				
Mother's Name					
First	✓	✓	✓	✓	
Middle	✓	✓	✓	✓	
Last	✓	✓	✓	✓	
Mother's Residence At Time of Pregnancy Outcome					
Street address		./			
City	v	∀ ✓			
County	,	y			
State	· •	· •			
Zip Code	✓	*			

Chapter 4 4-10 Data Variables

Table 4.2 Recommended Data Variables

Data Variable	Descriptive Epidemiology and Monitoring	Research	Service/ Planning	Linkage			
		nfant					
Text Description of Birth Defect	~	✓	✓				
Date of Death	✓	✓	✓	✓			
Birth Length	√	√	·				
Apgar Score	✓	✓					
Birth Order	✓	✓	✓				
Cytogenetic Analyses Performed	✓	✓	✓				
Diagnostic Tests and Procedures Performed	✓	✓	✓				
Autopsy Performed	✓	✓	✓				
Physicians of Record		✓	✓				
	М	other					
Date of Last Menstrual Period (LMP)	✓	✓					
Date of Ultrasound	✓	✓					
Gestational Age at Ultrasound	*	✓					
Mother's Medical Record Number(s)	✓	✓		✓			
Prenatal Diagnosis	✓	✓					
Mother's Social Security Number		✓		✓			
Census Tract of Maternal Residence at Pregnancy Outcome	~	✓		✓			
Mother's Telephone Number		✓	✓				
Mother's Education	✓	✓					
Prior Pregnancy History	✓	✓		✓			
Prenatal Care	✓	✓					
	Father						
Father's Date of Birth	✓	✓		✓			
Father's Name		√	✓				
Father's Education	√	√					
Father's Race	✓	√					
Father's Ethnicity	✓	√					
Father's Social Security #		✓		✓			

Chapter 4 4-11 Data Variables

4.9 References

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Chapter 4 4-13 Data Variables

Appendix 4.1 Descriptions of Minimum (Core) Data Variables

Appendix 4.1 Descriptions of Minimum (Core) Data Variables

Format for Variable Descriptions	A4.1-1
Infant	
Unique ID	A4.1-2
Date of Pregnancy Outcome	
Sex	A4.1-4
Infant's Name	
Source of Report	
Medical Record Number(s)	
Vital Record Certificate Number	
Place of Pregnancy Outcome.	
Pregnancy Outcome	
Birth Weight	
Plurality	A4.1-11
Gestational Age	
Diagnosis Code	A4.1-13
Contact Information	
Name of Responsible Party	A4.1-14
Address of Responsible Party	
Telephone Number of Responsible Party	A4.1-14
Mother	
Mother's Data of Birth	A4.1-15
Mother's Race	A4.1-16
Mother's Ethnicity	A4.1-16
Mother's Name	A4.1-17
Mother's Residence at Time of Pregnancy Outcome, Street Address	A4.1-18
Mother's Residence at Time of Pregnancy Outcome, City	A4.1-18
Mother's Residence at Time of Pregnancy Outcome, County	A4.1-19
Mother's Residence at Time of Pregnancy Outcome, State	A4.1-19
Mother's Residence at Time of Pregnancy Outcome, Zip Code	

Appendix 4.1 Data Variables

Appendix 4.1 Descriptions of Minimum (Core) Variables

Format for Variable Descriptions

Variable Name Name of data collection variable

Definition Definition of data collection variable

Justification Reason the birth defects program may want to include variable in

its database

Source Where variable comes from – abstracted, derived, created

Location Data sources and location within data sources where variable is

most likely to be consistently found

Type How variable should be stored – text, number, date, code (letters

and/or numbers), checkbox

Checks Any limits, ranges, or other criteria the variable should meet

Comments Other notes or comments about the variable

Options Recommended options for the variable

Variable Name Unique ID

Definition Identification code or number; a code or number that uniquely

identifies each case or record

Justification With a unique ID code, the birth defects program can refer to a

particular case more easily than having to refer to a set of other variables. For example, it is easier to refer to an abstract with ID 1234567 than to an abstract of John Doe, date of birth 04/27/1999,

born to mother Jane Doe.

The ID permits easy linkage between multiple data sets as long as each table contains the ID as one of its fields. This is essential for data transfer and processing, so that data for a particular case do not get mixed up with data from other cases. This field permits linking

multiple case reports for individual children.

Source Created by the registry as cases are added.

Location N/A **Type** Code

Checks Every individual in the database should have a *unique* ID.

Variable Name Definition Justification

Date of pregnancy outcome

Date of delivery or end of the index pregnancy

In conjunction with other fields, such as mother's last name, this field helps to identify a case uniquely. It is useful to researchers and social workers in verifying that they are referring to the pregnancy of interest when contacting mothers who may have had other pregnancies.

The birth defects program may require that, for live births, a diagnosis be made within a certain time period after the date of delivery (e.g., within one year) or by a particular age (e.g., prior to age 6). The date of delivery is necessary in order to determine whether the diagnosis was made within the time limit

Secular trends have been reported for certain birth defects (Nielsen et al., 2000; O'Leary et al., 1996; Centers for Disease Control and Prevention, 1992). The birth defects program can use the date of delivery in order to produce statistics and reports by delivery year and to examine secular trends in birth defects.

Cluster investigations are based on a defined diagnosis, geographical area, and time period. Knowing the delivery date allows investigators to determine which cases qualify to be included in a particular investigation.

Source Location

Abstracted

- Mother's delivery medical record (labor and delivery record)
- Infant's medical record (face sheet, labor and delivery record, birth certificate worksheet)
- Vital record

Type Checks

Date

Every record must have a pregnancy outcome date, except in cases of prenatal diagnosis where the pregnancy has not ended yet. The pregnancy outcome date should be after the mother's and father's date of birth, date of last menstrual period, and date of conception; on or after any prenatal diagnostic procedure date or prenatal ultrasound date; and on or before a postnatal procedure date.

Comments

Date of pregnancy outcome can be: date of birth, date of fetal demise, or date pregnancy ends.

Variable Name Sex

Definition Sex of the infant or fetus

Justification Birth defect risk may be associated with sex (Whiteman et al., 2000).

The birth defects program can use the sex of the infant or fetus in order

to evaluate differences in birth defect rates by sex.

Source Abstracted

Derived (from the karyotype)

Location • Mother's delivery medical record (labor and delivery record)

• Infant's medical record (face sheet, labor and delivery record, birth

certificate worksheet)

• Vital record

Type Code

Checks Every record should have sex recorded.

Comments If a karyotype was performed, the sex should match the karyotype,

except in rare cases of such discordances as XY females and XX males.

Options • Male

Female

Ambiguous

Unknown

Data Variables

Infant Variables – Core

Variable Name **Definition**

Justification

Infant's name

Name of infant or fetus

Field staff can use the infant's name and date of birth or pregnancy outcome date to locate medical records.

The birth defects program will use the name to unduplicate the reported cases and may employ the infant's name in addition to other fields to link to other data sets, such as vital records.

The infant's name is helpful when referring the family to social work, treatment, and prevention agencies.

Source Location Abstracted

Infant's medical record (face sheet, birth certificate worksheet)

Vital record

Type Checks

Comments

Text

If the infant's last name is the same as the father's or mother's last name, or a combination of the two, the spelling should usually match.

This variable may be collected as a single field or multiple fields. Separate fields for first, middle, last name, and suffix are recommended to improve unduplication and record linkage success. Individual fields of up to 25 digits each should be considered to avoid truncated names. This variable should include at least the infant's first and last name and may include the infant's middle name and any suffixes. An infant may be given more than one name or alias, sometimes referred to as "also known as" or AKA. The birth defects program may want to record all of the names, for easier linkage with other databases, to prevent duplication of cases in the database and to remain current with name use.

Fetuses resulting from fetal deaths and elective terminations often do not have names. The birth defects program should consider using the surname of the mother and inserting a standard first name (e.g., fetus) so that name data fields are complete in the database.

Variable Name Source of report

Definition Any data source where information was obtained or where a case report

originated.

Justification The source of report allows the birth defects program to identify where

information in a case abstract comes from. This is important for resolving data edit issues, for confirming the data, and for conducting

audits of facility reporting.

The data source fields permit the birth defects program to evaluate the

usefulness of utilizing specific facilities as data sources.

Source Abstracted

Location Any data source pertinent to program objectives **Type** Code, with allowance for multiple sources

Checks This field should always be filled out and should be a valid code.

Comments There can be multiple data sources for a given case. For example, an

infant may be identified with a birth defect at the delivery hospital, tertiary care hospital, cytogenetics laboratory, etc. (see also Chapter 6

on Case Ascertainment Methods).

Options It is useful to maintain a list of potential data sources and standard

codes (hospitals, clinics, laboratories, autopsy, etc.) unique to each

program.

Variable Name Medical record number(s)

Definition Medical record number(s) used by the source from which the

information was obtained.

Justification A medical record number allows facilities to retrieve records easily.

Although it may be possible to locate medical records using the patient's name and date of birth, the birth defects program may have a

name different than that recorded at the data source.

Source Abstracted

Location • Infant's medical record (face sheet)

Type Code

Comments Medical record numbers are not the same as visit, service, or encounter

numbers. Although not standard practice, multiple 'real' medical record numbers may be assigned to the same person, so it is important to identify each number for a given data source. Medical record numbers may also be very long. The birth defects program should make certain the computer program and registry database allow for entry of

the entire medical record number.

Variable Name Vital record certificate number

Definition Unique number assigned to a certificate by Vital Records.

Justification Vital record certificate numbers can be linked to other vital records

certificates. Often, vital records will reference or link the death

certificate to the birth certificate for infant deaths.

Programs can use an algorithm of data variables to find a potential match to a vital record. This process assists in identifying unique cases, establishing residency, and securing all of the data variables on the vital

record.

Birth certificate and fetal death certificate numbers can be the unique ID numbers for a program. Other 'program' numbers can be created using a similar format for cases that do not match to a birth certificate

or fetal death certificate.

Source Abstracted

Location On the certificate of birth, death, or fetal death and in the vital records

database

Type Number

Comments Separate fields for the live birth or fetal death and for the death record

number are recommended.

Variable Name Definition Justification

Place of pregnancy outcome

Location where the delivery or pregnancy outcome occurred Mother and infant records at the delivery facility often provide important information not found in tertiary care facility records (unless the delivery records are copied into the tertiary care records). The birth defects program can use the delivery location (hospital, midwifery, residence, etc.) to identify where delivery records need to be reviewed and abstracted.

The birth defects program may employ the delivery location in addition to other fields to link to other data sets, such as vital records.

The location where the delivery occurred allows the birth defects program to provide facility-specific statistics.

Source Location Abstracted

- Mother's delivery medical record (face sheet)
- Infant's delivery medical record (face sheet)
- Vital record

Type Checks

Comments

Code

This field should always be filled out and must be a valid code.

This includes those situations where delivery occurs outside of health care facilities as well as inside health care facilities.

Options

It is useful to maintain a list of potential data sources (hospitals, etc.) unique to each program.

- Home/residence
- Other
- Unknown

Variable Name

Pregnancy outcome

Definition

Outcome of the index pregnancy

Justification

The pregnancy outcome, in conjunction with gestational age fields, may determine whether a record should be included in the birth defects program.

At a minimum birth defect programs should distinguish the outcomes of live birth, fetal death, and induced termination.

Part of the mission of the birth defects program may be to refer families to social services. Since only live births would be referred to many of the services, it is important to know whether a given case is a live birth. Knowing which cases are elective terminations aids in evaluating trends in prenatal diagnosis, as well as evaluating the impact of prevention strategies such as folic acid supplementation and fortification.

Pregnancy outcome can be used to evaluate rates of birth defect by pregnancy outcome.

Source

Abstracted

Location

- Mother's delivery medical record (face sheet, discharge summary, labor and delivery record)
- Vital records

Type

Code

Checks

This field should always be filled out, except in cases of prenatal diagnosis where the pregnancy has not yet ended.

Comments

See Chapter 3 on Case Definition for definitions of pregnancy outcomes.

Variable Name

Birth weight

Definition

Weight of the infant or fetus at delivery

Justification

The birth weight may be needed for case definition if inclusion/exclusion criteria for selected birth defects, such as for

undescended testes and patent ductus arteriosus, are based on birth

weight.

In conjunction with gestational age, length, and head circumference, birth weight can be used to assess prenatal growth retardation, a

characteristic of fetal alcohol syndrome.

Source

Abstracted

Location

- Mother's delivery medical record (labor and delivery record)
- Infant's medical record (admission summary, labor and delivery record, birth certificate worksheet)
- Vital records

Type

Number

Checks

The birth weight must range between 0 and 10,000 grams or 0 and 50

pounds.

Comments

The data source may report birth weight in grams or kilograms, pounds and ounces, or pounds with decimals. The birth defects program may decide to record the weight in the units reported or in a uniform fashion, such as always as grams and kilograms. In this latter case, the birth defects program must be able to convert from one type of unit to another while collecting the data. Data fields can have computerized calculation functions.

Variable Name

Plurality

Definition

Number of fetuses or infants.

Justification

The plurality, in association with other fields such as county of residence and mother's social security number, can be used to avoid duplication of records in the birth defects program.

Knowing that the infant is from a multiple birth alerts the birth defects program that more effort may be needed to link to a particular vital record (Forrester and Canfield, 2000).

The birth defects program can use this data item to evaluate differences in birth defect rates for singletons and multiple births.

Source Location Abstracted

- Mother's delivery medical record (face sheet, admission summary, discharge summary, prenatal care record, labor and delivery record)
- Infant's medical record (face sheet, admission summary, discharge summary, prenatal care record, labor and delivery record, birth certificate worksheet).
- Vital records

Type

Number

Checks

This field should always be filled out.

Comments

Because some twin pregnancies are anomalous (for example, conjoined twins or fetus papyraceus), there may not be the expected two vital records for a pregnancy that is identified as a twin pregnancy.

Variable Name Gestational age

Definition Gestational age at pregnancy outcome

Justification Gestational age can be used to determine whether a pregnancy outcome

meets the case definition for the birth defects program.

Certain diagnoses are considered birth defects only when the infant is of a particular gestational age. For example, patent ductus arteriosus is common among premature infants and is often subject to exclusion

criteria before being counted as a birth defect.

Source Abstracted

Derived (see comments)

Location • Mother's delivery medical record (labor and delivery record)

• Infant's delivery medical record (admission summary, discharge summary, gestational age score record, labor and delivery record,

birth certificate worksheet)

Vital records

Type Number

Checks The gestational age should range between 0 and 52 weeks.

Comments The gestational age can be derived via several methods, and conflicting

gestational age information may be reported in the medical record (Alexander et al., 1990; Hall, 1990). As a result, the birth defects program will want to have a method for prioritizing gestational age

estimates from different sources.

Options See Chapter 3 on Case Definition for further information.

Infant Variables – Core

Variable Name Diagnosis code

Definition Code used for the diagnosis

Justification Coding birth defects eliminates the problem of having to sort through a

variety of differing descriptions. It allows for timely and efficient analyses of data and identification of cases for research and referral. Coding of birth defects enables birth defects researchers to know that

they are talking about the same birth defect, and allows for

comparability between different birth defects registries using the same

or comparable coding systems (Rasmussen et al., 2001).

SourceDerivedLocationN/ATypeCode

Checks Every case should have at least one diagnosis code (except if the birth

defects program includes non-malformed controls, in which case the program may create specific 'disease codes' for use as the data variable

in the diagnosis code field).

A case may have more than one diagnosis code. Every diagnosis

description should have a corresponding code and vice versa.

Comments The International Classification of Disease (ICD) coding system is the

standard used in the health care delivery system. NBDPN currently requires that programs report cases using ICD-9-CM codes. The registry should accommodate a minimum of 15 unique diagnostic

codes per case.

Options The recommended coding system is the CDC 6-digit code, which is

easily converted to ICD-9-CM. See the Chapter 5 on Classification and

Coding for further information.

Contact Information – Core

Variable Name Name of responsible party

Definition Name of parent, custodial parent, or guardian

Justification Useful in programs that refer a family to services when contact with a

parent may be inappropriate.

Source Abstracted

Location • Face sheet, signed authorization, social worker's notes, birth

certificate worksheet

• Immunization registry, metabolic screening database

Vital record

Type Text

Comments The name may be collected as a single field or multiple fields for first,

middle, and last name. Allowing for up to 25 characters for each

portion of the name should be considered.

Variable Name Address of responsible party

Definition Address of parent, custodial parent, or guardian

Justification Useful in programs that refer a family to services when contact with a

parent may be inappropriate.

Source Abstracted

Location • Face sheet, signed authorization, social worker's notes, birth

certificate worksheet

• Immunization registry, metabolic screening database

Vital record

Type Text

Comments Include fields for the street address, city, state, and zip code. Allow at

least 25 digits for street address and 20 digits for city name. If the residence address and the mailing address of the responsible party are

different, collect the mailing address for this item.

Variable Name Telephone number of responsible party

Definition Telephone number of child's parent, custodial parent, or guardian

Justification Useful in programs that refer a family to services when contact with a

parent may be inappropriate and when telephone contact may be

indicated.

Source Abstracted

Location • Face sheet, signed authorization, social worker's notes, birth

certificate worksheet

• Immunization registry, metabolic screening database

Vital record

Type Number

Comments Include area code with number.

Variable Name Definition Justification

Mother's date of birth

Birth mother's date of birth

In conjunction with other fields, such as mother's name, the birth defects program field staff can use the mother's date of birth to locate medical records when the mother's medical record number is not known. The birth defects program can use the mother's date of birth and other fields to determine whether a case has been abstracted or added to the registry under a different ID.

The birth defects program can employ the mother's date of birth in addition to other fields to link to other data sets, such as vital records or Medicaid.

The birth defects program can use the mother's date of birth and infant's date of delivery in order to calculate the mother's age at delivery. The mother's age at delivery can then be used in clinical review.

Source Location

Abstracted

- Mother's delivery medical record (face sheet, prenatal care record)
- Infant's medical record (face sheet, prenatal care record, birth certificate worksheet)
- Vital records

Type

Date

Checks

This date must be earlier than all other dates except possibly the father's date of birth. Medical records may sometimes confuse maternal and paternal information. If the mother's date of birth is the same as the father's date of birth, the birth defects program should double check to make certain that this is true.

Comments

See also Chapter 6 on Case Ascertainment Methods, the section on Data Sources.

Variable Name Mother's race **Definition** Birth mother's race

Justification The birth defects program can use the mother's race in order to present

data on birth defect rates by maternal race, one of the most important

person variables in descriptive epidemiology.

Abstracted Source

Location Mother's delivery medical record (face sheet, prenatal care record)

Infant's medical record (admission summary, prenatal care record,

birth certificate worksheet) Vital records

Code **Type**

Comments Racial categories and codes used by birth defects surveillance programs

should be compatible with the federal standards in current use for race.

Mother's ethnicity Variable Name **Definition** Birth mother's ethnicity

Ethnicity is a designation separate from maternal race. The birth defects Justification

program can use the mother's ethnicity in order to evaluate differences

in birth defect rates by maternal ethnicity.

Source Abstracted

Location Mother's delivery medical record (face sheet, prenatal care record)

> Infant's medical record (admission summary, prenatal care record, birth certificate worksheet)

Vital records

Type Code

Comments

It is important to collect ethnicity data that meets the needs of the registry to monitor the health of the ethnic populations within the state. Generally, each state Department of Public Health will have identified the populations of special interest to that state. Ethnicity categories and codes should be compatible with the federal standards in current use

for ethnicity.

Variable Name Definition Justification

Mother's name

Full name of birth mother

In conjunction with other fields, such as mother's date of birth, the birth defects program field staff can use the mother's name to locate medical records when the mother's medical record number is not known. The birth defects program can employ the mother's name in addition to other fields to unduplicate case reports and to link to other data sets, such as vital records or Medicaid.

The mother's name is needed so that she can be contacted by researchers conducting approved studies and by social workers or others for outreach efforts.

Source Location

Abstracted

- Mother's delivery medical record (face sheet)
- Infant's medical record (face sheet, birth certificate worksheet)
- Vital records

Type Comments

Text

This variable may be collected as a single field or multiple fields. This variable should include at least the mother's first and last name and may include the mother's middle name and maiden name (name before marriage). A woman may have more than one name or alias (also known as or AKA). Separate fields for first, middle, and last name and for maiden name are recommended. Field lengths of 25 characters or larger for each portion of the name should be considered. The birth defects program should record all of the names, for easier linkage with other databases and to prevent entering duplicate cases in the database.

Variable Name Definition Mother's street address of residence at pregnancy outcome

Street address of birth mother's residence at the time of the outcome of

the index pregnancy

Justification Street address is necessary for geocoding location of residence and

linking with other data systems through geographical information systems (GIS). The street address field may be needed when assigning the county of residence, particularly when a city includes part of more

than one adjacent county.

Source

Abstracted

Location

- Mother's delivery medical record (face sheet)
- Infant's delivery medical record (face sheet, birth certificate worksheet)
- Vital records

Type

Text

Comments Include apartment numbers, etc. A field length of up to 40 characters

should be considered.

If there is a difference between residence address and mailing address, choose residence address. Only use P.O. Box if there is no physical address for the mother

Variable Name Definition

Mother's city of residence at pregnancy outcome

City of address of birth mother's residence at the time of the outcome

of the index pregnancy

Justification The city at delivery field is often needed when assigning the county of

residence.

Source Abstracted

Derived (from zip code or census tract number)

Location • Mother's delivery medical record (face sheet)

• Infant's delivery medical record (face sheet, birth certificate

worksheet)

Vital records

Type

Text

Comments

If there is a difference between residence address and mailing address, choose residence address. Allow for up to 25 characters for city name text fields. A separate city code field may be used to correspond with the city name to facilitate statistical analysis. City coding structures should be compatible with Federal Information Processing Standards (FIPS).

Variable Name

Mother's county of residence at pregnancy outcome

Definition

County code of birth mother's county of residence at the time of the

outcome of the index pregnancy

Justification

The county of residence, in association with other fields such as plurality and mother's social security number, can be used to avoid

duplication of records in the registry.

Source

Abstracted

Derived (from street address and city, zip code, or census tract number)

Location

- Mother's delivery medical record (face sheet)
- Infant's delivery medical record (face sheet, birth certificate worksheet)
- Vital records

Type

Code

Comments

If there is a difference between residence address and mailing address, choose residence address. County coding schemes should be compatible with standard federal FIPS codes.

Variable Name Definition

Mother's state of residence at pregnancy outcome

State in which birth mother resided at the time of the outcome of the

index pregnancy

Justification

The state in which the mother resided is needed if the birth defects program's inclusion criteria include only residents of a certain state. The state of residence, along with other address components, is needed so that researchers and social workers can contact the family, provided a more recent address is not known.

Source

Abstracted

Derived (from other residence information such as city, zip code, and census tract number)

Location

- Mother's delivery medical record (face sheet)
- Infant's delivery medical record (face sheet, birth certificate worksheet)
- Vital records

Type

Comments

Standard 2-letter state codes used by US Postal Service

If there is a difference between residence address and mailing address, choose residence address. Procedures for reporting information for places outside the US need to be contemplated.

Appendix 4.1 A4.1-19 Data Variables

Variable Name Definition Mother's zip code of residence at time of pregnancy outcome

Zip code of birth mother's residence at the time of the outcome of the

index pregnancy

Justification Cluster investigations are based on a defined diagnosis, geographical

area, and time period. Knowing the zip code of residence may allow investigators to determine which cases qualify to be included in cluster

investigations.

The zip code, along with other address components, is needed so that researchers, social workers, and others can contact the family, provided

a more recent address is not known.

Source Abstracted

Derived (from other residence information such as street address and

city or census tract number)

Location • Mother's delivery medical record (face sheet)

• Infant's delivery medical record (face sheet, birth certificate worksheet)

Vital records

Type

Number

Comments

If there is a difference between residence address and mailing address, choose residence address. This code only applies to United States zip codes and may be the 5-digit or the 9-digit code.

Appendix 4.2

Descriptions of Recommended Data Variables

Appendix 4.2 Descriptions of Recommended Data Variables

Format for Variable Descriptions	A4.2-1
Infant	
Text Description of Birth Defect	A4.2-2
Date of Death	A4.2-3
Birth Length	A4.2-3
Apgar Score	A4.2-4
Birth Order	A4.2-4
Cytogenetic Analyses Performed	A4.2-5
Diagnostic Tests and Procedures Performed	A4.2-6
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Physicians of Record	A4.2-8
Mother	
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Data of Ultrasound	A4.2-10
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Father	
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Father's Education	A4.2-17
Father's Race	A4.2-17
Father's Ethnicity	A4.2-18
Father's Social Security Number	A4.2-18

Appendix 4.2 Descriptions of Recommended Data Variables

Format for Variable Descriptions

Variable Name Name of data collection variable

Definition Definition of data collection variable

Justification Reason the birth defects program may want to include variable in

its database

Source Where variable comes from – abstracted, derived, created

Location Data sources and location within data sources where variable is

most likely to be consistently found

Type How variable should be stored – text, number, date, code (letters

and/or numbers), checkbox

Checks Any limits, ranges, or other criteria the variable should meet

Comments Other notes or comments about the variable

Options Recommended options for the variable

Variable Name Definition Justification

Text description of birth defect

Description of diagnosis

A birth defect may be diagnosed based on more than one procedure or examination. Moreover, two procedures or clinicians may provide different details about the birth defect. For example, one procedure may report that the infant had a myelomeningocele, while a second may mention a lumbar spina bifida. These should all be combined into a single description such as lumbar myelomeningocele. Or one procedure may mention the infant had a cleft lip and palate, while a second notes that the cleft lip was only on the left side of the mouth. These should be combined into something like left cleft lip and palate.

The birth defect description recorded in text format in the data makes it easier to assign disease codes when the medical record is no longer available.

Source Location

Abstracted

- Mother's delivery medical record (prenatal care record, labor and delivery record, prenatal diagnostic procedure reports)
- Infant's medical record (face sheet, admission summary, discharge summary, procedure reports, consultation reports, labor and delivery record, birth certificate worksheet)
- Vital records

Type

Text

Checks

Every case should have at least one diagnosis description (unless the birth defects program includes non-malformed controls). A case may have more than one diagnosis description. Every diagnosis description should have a corresponding code and vice versa.

Comments

See Chapter 5 on Classification and Coding.

Appendix 4.2 A4.2-2 Data Variables

Variable Name **Definition**

Date of death

Justification

The date when the death occurred

The date of death permits the birth defects program to know that most postnatal procedures will not occur after this date, the exceptions being such procedures as autopsies, cytogenetic analyses, and other laboratory analyses.

The delivery date for a live birth along with the date of death can be used to determine length of survival. Researchers can use this to calculate survival rates for specific lengths of time.

Source

Abstracted

Location

- Mother's delivery medical record (prenatal care record, labor and delivery record, prenatal diagnostic procedure reports)
- Infant's medical record (face sheet, admission summary, discharge summary, procedure reports, consultation reports, labor and delivery record)
- Vital records

Type

Date

Checks

This field should only be filled out if the pregnancy outcome is "live birth". The date of death should be on or after the date of delivery.

Variable Name

Birth Length

Definition

Length of newborn at birth

Justification

In conjunction with gestational age, birth weight, and head circumference, length can be used to assess prenatal growth retardation,

a characteristic of fetal alcohol syndrome. However, these

circumstances account for only a small subset of cases a birth defects program will collect, and it may not be worth collecting the information

on all cases.

Source

Abstracted

Location

Infant's medical record (labor and delivery record)

Vital records in some states

Type

Number

Checks

Edit checks for range and for consistency with gestational age are

recommended.

Options

Best collected as centimeters.

Variable Name

Apgar score

Definition

Clinical assessment score of newborn at delivery

Justification

Apgar scores are a gross measure of early neonatal health. If the scores are low, that means that the newborn had cardiorespiratory problems immediately after delivery. These problems may or may not be related to a birth defect in the infant. For example, some postnatal complications that correlate with low Apgar scores (intestinal perforations, intraventricular hemorrhage) overlap with problems caused by birth defects.

Source

Abstracted

Location

- Infant's medical record (labor and delivery record), birth certificate work sheet.
- Vital records

Type

Code

Checks

Values from 0 through 10 or coded unknown/not applicable.

Comments

Apgar scores at 1 minute, 5 minutes, and 10 minutes are often available. Vital records generally provide 1- and 5-minute scores, with a change to 5- and 10-minute scores for low 5-minute scores being

implemented nationwide.

Variable Name

Birth order

Definition

Order of delivery for multiple births.

Justification

Birth order is the order in which infants of a multiple gestation

pregnancy are delivered.

In cases of multiple gestation pregnancies, delivery records might not refer to the infants or fetuses by name but by some other designation such as Twin A and Twin B. This might make it difficult to determine which vital records a particular infant or fetus should be linked to. Vital records may record birth order. Thus birth order might be useful for linkage with vital records in cases of multiple gestation pregnancies. However, other variables such as infant or fetus sex and birth weight might prove as useful for linkage.

Source

Abstracted

Location

- Infant's medical record (labor and delivery record), birth certificate worksheet.
- Vital records

Type

Number

Checks

Must be less than or equal to plurality.

Variable Name Definition Justification

Cytogenetic analyses performed

Whether or not a cytogenetics analysis was performed.

Certain structural birth defects are associated with chromosomal abnormalities (Boudjemline et al., 2001; Bullen et al., 2001; Torfs and Christianson, 1998). Structural defects in the presence of a chromosomal abnormality are often considered to be secondary to or the result of the chromosomal abnormality. Such cases may not be considered suitable for research into potential environmental causes of structural defects. And analyses of the proportion of structural defects associated with chromosomal abnormalities often are based on the number of cases where the karyotype is known, because some of the cases without a chromosome analysis may be expected to have chromosomal abnormalities. Infants with certain chromosomal abnormalities also have higher mortality and morbidity than infants without chromosomal abnormalities. Thus it may be important to know whether a chromosomal abnormality is present when deciding whether to refer cases for intervention or prevention activities.

It is also important to know whether a chromosome analysis was performed at all, even if the results of the analysis are not in the medical record. If time and resources are available, and the chromosome analysis results are considered important, the birth defects program may attempt to track down the results of the analysis. Knowledge that a chromosome analysis was not performed (e.g., because the parents refused) or that a chromosome analysis failed will prevent a birth defects program from wasting resources searching for chromosome analysis results that do not exist.

Source Location Abstracted

 Infant's medical record (face sheet, admission summary, discharge summary, procedure reports, consultation reports, labor and delivery record)

Type Checks Number/Code

Must be yes, no, or unknown.

Variable Name Definition Justification

Diagnostic tests and procedures performed

Method used to reach diagnosis.

Different procedures can be used in the diagnosis of a birth defect. Moreover, procedures differ in their accuracy and reliability in diagnosing certain birth defects. For example, Down syndrome can usually be considered to be more definite when it is based on a chromosome analysis than on a physical examination of the infant/fetus. Thus it is often not enough to know that a birth defect was mentioned in a medical record; it is important to know how the birth defect diagnosis was made. Moreover, the researcher may only be interested in birth defects identified by particular procedures. For example, researchers may only be interested in cases of a heart defect identified through fetal echocardiography.

If a birth defects program has clearly defined case inclusion criteria (e.g., infants and fetuses with certain birth defects are only included if diagnoses were made by certain procedures), then basic research can be conducted. An example would be for a birth defects program to only include cardiac defects diagnosed by echocardiography, cardiac catheterization, prenatal ultrasound, or autopsy.

Source Location

Abstracted

 Infant's medical record (face sheet, admission summary, discharge summary, procedure reports, consultation reports, labor and delivery record)

Type Checks Comments

Code

Any limits, ranges, or other criteria the data variable should meet. Must develop an appropriate coding structure or select a coding standard such as CPT coding to aid in capturing and tabulating the information.

Options

May collect primary diagnostic method using a specific hierarchy based on diagnostic accuracy or include multiple procedures fields.

Variable Name Definition Justification

Autopsy performed

Indicates whether an autopsy was conducted.

The autopsy is considered one of the more definitive procedures for identifying structural birth defects.

However, even if an autopsy is performed, the autopsy information is not always added to the medical record. As long as a birth defects program has clearly defined case inclusion criteria (e.g., infants and fetuses with certain birth defects are only included if diagnoses were made by certain procedures), then basic research can be conducted. An example of such inclusion criteria would be for a birth defects program to only include cardiac defects diagnosed by echocardiography, cardiac catheterization, prenatal ultrasound, or autopsy.

Source

Abstracted

Location

- Infant's medical record (face sheet, admission summary, discharge summary, procedure reports, consultation reports, labor and delivery record)
- Death certificate, fetal death report.

Type

Code

Checks

Not applicable for live births still living.

Options

Values would include yes, no, or unknown/not applicable.

Variable Name Physicians of record

Definition Physician(s) identified as being responsible for admission and

discharge records.

Justification A birth defects program might want to have information on the

physicians of record in order to obtain additional information, to determine if all appropriate referrals were made, to alert physician to need for folic acid recommendations, or to obtain permission to contact

the family.

Source Abstracted

Location

• Infant's medical record (face sheet, admission summary, discharge summary, procedure reports, consultation reports, labor and

delivery record)

• Newborn metabolic screening data

Vital record

Type Data are stored as text (names and addresses)

Checks N/A

Comments

To be useful, this information should include name and address for the

physician. Allow for 40 characters for entry of each name. There may be interest in collecting multiple physicians and their role, as in pediatrician, obstetrician, or family practice physician to clarify

appropriate physician depending upon circumstance.

Appendix 4.2 A4.2-8 Data Variables

Variable Name Date of last menstrual period (LMP)

Definition First day of last menstrual period

Justification Date of LMP, along with date of delivery, can be used to calculate

gestational age at delivery. Gestational age at delivery can be used for determining if a spontaneous fetal death or pregnancy termination

meets the case definition for the registry.

Source Abstracted

Derived (see comments)

• Mother's delivery medical record (prenatal care record, labor and

delivery record)

• Infant's delivery medical record (prenatal care record, labor and

delivery record)

Vital records

Type Date

Checks The LMP date must be before the date of delivery, estimated date of

delivery, prenatal ultrasound date, and prenatal and postnatal procedure dates. The LMP date should not be more than one year before the date of delivery, estimated date of delivery, prenatal ultrasound date, and

prenatal procedure dates.

Comments If the LMP date is recorded in both the prenatal records and the

admission interview, use the LMP date in the prenatal records. If more than one LMP date is found in the prenatal records, record the earliest

LMP date in this field.

Options See Chapter 3 on Case Definition for further information.

Variable Name Date of ultrasound

DefinitionDate of the earliest identified ultrasound used to assess gestational age **Justification**Date of ultrasound, along with gestational age at time of ultrasound and

delivery date, can be used to calculate gestational age at delivery.

Source Abstracted

• Mother's delivery medical record (prenatal care record, labor and

delivery record)

• Infant's delivery medical record (prenatal care record, labor and

delivery record)

Type Date

Checks The date of ultrasound field should only be filled in when the

gestational age at ultrasound is also known. The ultrasound date must be before or on delivery date and postnatal procedure dates and after the LMP date. The ultrasound date should not be more than 10 months

before the date of delivery.

Comments Only record information present in the medical record. DO NOT

calculate gestational ages or dates.

If multiple ultrasounds were done to determine gestational age, record

the date of the earliest ultrasound.

Variable Name

Gestational age at time of ultrasound

Definition

Gestational age (in weeks) at the time of ultrasound, as estimated by the

earliest ultrasound performed

Justification

Gestational age at ultrasound combined with date of pregnancy outcome can be used for determining if a spontaneous fetal death or pregnancy termination meets the case definition for the registry.

Certain diagnoses are considered birth defects only when the infant is of a particular gestational age. For example, patent ductus arteriosus is common among premature infants and is only considered a birth defect if found in infants born at term.

Abstracted

Source Location

 Mother's delivery medical record (prenatal care record, labor and delivery record)

• Infant's delivery medical record (prenatal care record, labor and delivery record)

Type

Number

Checks

This field should only be filled in when the date of ultrasound is also known. The gestational age at ultrasound should range between five menstrual weeks and birth.

Comments

Only record information present in the medical record. Do not calculate gestational ages or dates.

If multiple ultrasounds were done to determine gestational age, record the date of the earliest ultrasound.

Variable Name

Mother's medical record number(s)

Definition

Birth mother's medical record number(s)

Justification

A medical record number allows facilities to retrieve records more

easily.

Source

Abstracted

Location

• Mother's delivery medical record (face sheet)

Type

Code

Comments

The mother may have more than one medical record at a given hospital. Medical record numbers may also be very long. Allow for up to 12 alphanumeric characters for this field. The birth defects program should make certain the computer program allows for entry of the entire medical record number.

Variable Name Definition

Prenatal diagnosis

The diagnosis made before birth by prenatal diagnostic procedures and tests and neither confirmed nor ruled out by postnatal procedures and tests

Justification

Prenatal diagnostic procedures used to detect structural birth defects may not be considered to support as definitive a diagnosis as postnatal procedures, and prenatal detection of a birth defect is frequently considered to be tentative. Often physicians will attempt to verify or refine the prenatal diagnosis postnatally, such as through physical examinations, x-rays, or ultrasounds of the live birth or through autopsy of fetal deaths and elective terminations. Thus birth defects program staff should determine whether postnatal procedures and tests were performed and the results of such procedures and tests. However, postnatal confirmation or clarification of prenatally detected birth defects may not always be possible. In such cases the diagnoses identified through prenatal diagnostic procedures and tests are the best information available. Thus it may be useful for the birth defects program to indicate those diagnoses based solely on prenatal procedures.

Source Location Abstracted

- Mother's delivery medical record (prenatal care record, labor and delivery record, prenatal diagnostic procedure reports)
- Infant's medical record (face sheet, admission summary, discharge summary, procedure reports, consultation reports, labor and delivery record, birth certificate worksheet)
- Vital records

Type Comments

Checkbox

Prenatal cytogenetic tests may also be considered suspect. Depending on the source of the cell sample used, the sample could have been contaminated by maternal cells. Or, as in the case of chorionic villus sampling, any chromosomal abnormalities identified may be limited to the source of the cell sample and may not affect the fetus. However, prenatal cytogenetic tests are usually considered to be of greater validity than prenatal procedures for identifying structural defects.

Variable Name Mother's Social Security number

Definition Birth mother's Social Security number

Justification The mother's Social Security number, in association with other fields

such as plurality and county of residence, can be used to avoid

duplication of records in the registry.

The birth defects program can employ the mother's Social Security number to link to other data sets, such as the Medicaid database.

Source Abstracted

Location • Mother's delivery medical record (face sheet, prenatal care record)

• Infant's medical record (face sheet, prenatal care record, birth

certificate worksheet)

Vital records

Type Number

Checks Medical records may sometimes confuse maternal and paternal

information. The mother's and father's Social Security numbers should

not be the same.

Variable Name

Census tract of maternal residence at pregnancy outcome

Definition Census tract number of birth mother's residence at the time of the

outcome of the index pregnancy

Justification The geographical areas in most cluster investigations to date have been

counties, cities, or particular zip codes. However, in the future, cluster and other investigations may focus on geographical areas defined in other ways. Knowing the census tract number at delivery may allow investigators to determine which cases qualify to be included in such

future investigations.

Source Derived

Abstracted (from vital records files)

Location • Vital records

Type Number

Variable Name Mother's telephone number

Definition Birth mother's most recent telephone number: area code and telephone

number

Justification The mother's telephone number is needed so that researchers and social

workers can contact the family.

Source Abstracted

Location • Mother's delivery medical record (face sheet)

Infant's medical record (face sheet)

Type Number

Comments Enter the area code and seven-digit telephone number. If the area code

is not known, enter only the seven-digit telephone number. Note that the telephone number found in a tertiary care facility is more likely to

be current than the telephone number at the birth hospital.

Variable Name Mother's education

Definition Birth mother's highest level of education attained

Justification Education can be used as an indicator of socioeconomic status (SES).

Collecting maternal education would allow the birth defects program to

evaluate its relationship to birth defect risk.

Source Abstracted

Location • Birth certificate worksheet

• Birth certificate, fetal death report

Type Code

Checks Any limits, ranges, or other criteria the data variable should meet.

Comments Since maternal education is not reported consistently in medical

records, this information can be obtained more easily by linking to vital

record certificates.

Options Method for storing the information should permit identifying cases

with less than high school, high school, some college, and college

graduate.

Variable Name Prior pregnancy history

Definition Prior live births and fetal deaths to the birth mother

Justification Information can be used to identify women with a significant history of

fetal loss or infant death.

Source Abstracted

Location • Mother's delivery medical record (prenatal care record, labor and

delivery record, prenatal diagnostic procedure reports)

Type Number

Comments This information reflects the number of prior live births and fetal

deaths the mother has experienced. Vital record would provide prior live births now living and prior live births now deceased. Medical

record can provide parity and gravida.

Variable Name Definition

Justification

Prenatal care

Information on the initiation and extent of prenatal care

Data on prenatal care (such as month of prenatal care and number of prenatal visits), may be useful to a birth defects program. Knowing that the mother did or did not have prenatal care may be useful for birth defects program staff in evaluating other fields on the form. E.g., if it is known that the mother did not have prenatal care, there is less likelihood of finding information on prenatal tests or mother's medical

history. And prenatal care may be used as an indication of other factors

such as socioeconomic status (SES).

However, birth defects usually occur before pregnancy is recognized and prenatal care can begin. Furthermore, prenatal care may not be consistently or accurately reported in the medical record – the mother may move or change health care providers or the prenatal care visit information may not be counted consistently. There may be differences

of opinion as to what qualifies as a prenatal visit.

Source Abstracted Derived

Mother's delivery medical record (prenatal care record, labor and delivery record, prenatal diagnostic procedure reports)

derivery record, prenatar diagnostic procedure reports)

Type Number

Checks Range checks and consistency with woman's age

Comments The information to be considered for inclusion would be month

prenatal care began and number of prenatal visits.

Options The prenatal care information can be summarized using the Kotelchuck

or possibly the Kessner Index to standardize the information for more

meaningful analysis.

Variable Name Definition

Justification

Father's date of birth

Date of birth for father

The birth defects program may employ the father's date of birth in addition to other fields to link to other data sets, such as Medicaid. Paternal age may be associated with risk for certain birth defects (McIntosh et al., 1995; Olshan et al., 1994). The information can be useful in studies of paternal occupational or exposure cohort studies into associations with birth defects in progeny.

The birth defects program can use the father's date of birth and infant's date of delivery in order to calculate the father's age at delivery. The father's age at delivery can then be used in analyzing birth defect rates by paternal age.

Source

Abstracted

Location

- Medical record, birth certificate worksheet
- Birth/fetal death, death record

Type

Text

Checks

Range checks for father's ages under 12.

Variable Name

Father's name

Definition

Name of father

Justification

The birth defects program may employ the father's name in addition to other fields to link to other data sets, such as vital records or Medicaid.

However, information on the birth father is not consistently found in medical records or vital records.

Source

Abstracted

Location

- Medical record and birth record worksheet
- Birth/fetal death and death record

Type

Text

Comments

The name may be a single field or may be stored as separate first, middle, last, and surname suffix fields. Separate fields greatly facilitate record linkage. Providing 25 character fields for first, middle, and last names should be considered.

Variable Name Father's education

Definition Father's highest level of education attained

Justification Socioeconomic status (SES) can influence risk of having an infant with

a birth defect. Collecting paternal education would allow the birth

defects program to evaluate its impact on birth defect risk.

Source Abstracted

Location • Birth certificate worksheet

Birth/fetal death record

Type Code

Checks Consistency between father's age and education

Comments Since paternal education is not reported consistently in medical records,

this information can be obtained more easily by linking to vital record

certificates.

Options Method for storing the information should permit identifying cases

with less than high school, high school, some college, and college

graduate.

Variable Name Father's race

Definition Race of father

Justification The birth defects program can use the birth father's race in order to

evaluate differences in birth defect rates and examine program goals

and activities by paternal race.

Source Abstracted

Location • Birth certificate worksheet

Birth/fetal death record

Type Code

Comments Racial categories and codes used by birth defects surveillance programs

should be compatible with the federal standards in current use for race.

Variable NameFather's ethnicityDefinitionEthnicity of father

Justification Ethnicity is a designation separate from race. The birth defects program

can use the father's ethnicity in order to evaluate differences in birth defect rates or outreach effort goals and activities by father's ethnicity.

Source Abstracted

Location • Birth certificate worksheet

Birth/fetal death record

Type Code

Checks Should be valid code.

Comments Must develop a code structure that meets registry needs and reflects

available data on ethnicity. Should be compatible with federal standard

for ethnicity classification.

Variable Name Father's Social Security number

Definition Social Security number of the father

Justification The birth defects program can employ the father's Social Security

number to link to other data sets, such as the Medicaid database.

Source Abstracted

Location • Medical record, birth certificate worksheet

• Birth/fetal death record

Type Number

Checks Medical records may sometimes confuse maternal and paternal

information. The mother's and father's Social Security numbers should

not be the same

Chapter 5 Classification and Coding

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^{*}This document may be viewed or downloaded from the NBDPN website at: http://www.nbdpn.org/bdsurveillance.html

5.1 Introduction

The National Birth Defects Prevention Network (NBDPN) promotes the use of coded information that is comparable across birth defects programs and methods of case ascertainment, especially for conditions that are reported annually to NBDPN. The proper and accurate coding of diagnostic information is an essential aspect of birth defects surveillance.

A disease classification system plays an important role in the ability of surveillance systems to collect, code, retrieve, and translate information regarding diagnoses and procedures. These activities depend on the ability to assign specific codes to medical information, based on a standardized classification scheme. There are two important ways that classification systems and the coding of birth defects within those systems are central to the surveillance process. Classification and coding rely on a standardized set of rules and procedures for case ascertainment based on medical information, as well as on a standardized way of describing and organizing "cases" based on their clinical conditions.

Coded medical information has become an important part of the health care delivery system. Coding rules, guidelines, and standards have evolved for practically every type of health service encounter. Surveillance systems should understand the various factors that affect the quality of the coding of birth defects and should implement procedures to improve the utility of coding.

In this chapter we discuss disease classification systems (Section 5.2), classification issues that affect surveillance systems (Section 5.3), guidelines for effective coding (Section 5.4), quality issues related to coded data (Section 5.5), and tips and hints to assist with the classification and coding aspects of managing a surveillance system (Section 5.6). References cited in this chapter may be found in Section 5.7.

The two appendices to this document may be viewed or downloaded from the NBDPN website at http://www.nbdpn.org/bdsurveillance.html. Appendix 5.1 is the Texas Disease Index and Appendix 5.2 is the listing of CDC 6-digit codes.

5.2 Disease Classification Systems

Over time, a number of systems for classifying pathology, diseases, injuries, and clinical procedures have been developed. This has led to a classification system known as the International Classification of Diseases (ICD). At present, the World Health Organization (WHO) and 10 international centers coordinate classification efforts and promote a standardized classification system for organizing coded data for storage, retrieval, and analysis. Using a standardized system, disease information that is collected by various medical professionals can be compared, grouped, and tabulated for statistical purposes. Definitive information about disease classification in the United States is available from the National Center for Health Statistics (NCHS) of the Centers for Disease Control and Prevention (CDC) (see http://www.cdc.gov/nchs/icd9.htm).

The ninth revision of the International Classification of Diseases (ICD-9) was in use from 1979 to 1998. The United States uses the standard ICD version for coding deaths and in 1979 developed a 'clinical modification' for use in hospitals (i.e., ICD-9-CM). The clinical modification of the ICD-9 expanded the general categories, permitted greater detail and description, and included codes for clinical procedures. A large body of ICD-CM coding guidelines and rules was also developed. Since most of the information about birth defects comes from clinical records, the discussion below refers primarily to ICD-9-CM. However, comments regarding structure and organization are applicable to ICD-9 codes as well.

In 1999, the tenth revision (ICD-10) became operational for coding causes of death on death certificates. Although the classification structure is basically unchanged, ICD-10 reflects a significant revision from ICD-9. The codes are alphanumeric instead of numeric (as was the case in ICD-9), there are more general categories, and the codes are described in greater detail than in earlier versions. As of 2002, NCHS is developing the clinical modification ICD-10-CM. (This reference manual will not discuss ICD-10-CM until it is implemented).

Of importance to birth defects surveillance is the fact that, although ICD-9-CM is an acknowledged standard for coding medical information, it is not optimal for the level of detail required for coding many birth defects.

In 1979 the British Paediatric Association (BPA) developed a classification of diseases by modifying ICD-9-CM (British). In 1983, staff in CDC's birth defects branch modified the BPA coding system and developed a classification system specific to birth defects coding. The 6-digit CDC code is a classification system that allows coding of more detailed descriptions of birth defects and related conditions (see Appendix 5.2 for a complete listing of the 6-digit CDC codes).

5.2.1 Description and Format

The ICD-9-CM and the 6-digit CDC coding systems are divided into general categories that include body systems, medical conditions, and other health-related issues. The codes are hierarchical and expand to reflect specific conditions within a general category. Each code category is populated with specific diseases and related conditions. In ICD-9-CM the majority of the codes used in birth defects programs is between the code categories 740 and 759, which come under the general heading of 'congenital anomalies'. The ICD-9-CM and the 6-digit CDC coding systems utilize a similar format for categorizing disease. ICD-9-CM utilizes up to five digits, while the CDC coding system utilizes six.

5.2.2 ICD-9-CM and the 6-digit CDC Code – A Comparison

In most cases, the first four digits of the 6-digit CDC code are identical to the first four digits of the ICD-9-CM code. This enables birth defects programs to utilize the coded data collected from hospital data sets, while at the same enhancing the level of coding detail for birth defects program use. Since, the 6-digit CDC code usually collapses into the ICD-9-CM at the fourth-digit level, programs that use ICD-9-CM codes have data that are comparable between states.

The most significant difference between ICD-9-CM and the 6-digit CDC code is reflected in the level of detail indicated by the sixth digit. The sixth digit can be used to indicate one of three aspects of the defect:

- > Laterality of the defect
 - .001 Left side only
 - .002 Right side only
 - .003 Unilateral, unknown which side
 - .004 Bilateral; both sides
- > Greater specificity for a particular defect
 - .005 Example: 756.615 Diaphragmatic hernia (Bochdalek)
 - .006 Example: 756.616 Diaphragmatic hernia (Morgagni)
 - .007 Example: 756.617 Hemidiaphragm
- Incomplete confirmation of a defect (includes possible or probable or only diagnosed prenatally)
 .008 Example: 745.498 Probable Ventricular Septal Defect (VSD)

5.3 Classification Issues That Affect Surveillance Systems

It is important to recognize that there are advantages and disadvantages associated with both the ICD-9-CM and 6-digit CDC coding systems. Programs need to be aware of the ways in which these may affect data quality and other surveillance activities.

- Any coding system is limited to the number of literal descriptions assigned to a code in the system. In other words, there may be synonyms for one birth defect, or many related birth defects may be assigned to one code. The disease index provided in Appendix 5.1 is a tool that can be used to assist with coding. This alphabetic cross-linked index of birth defects and corresponding 6-digit CDC codes was developed by the Texas Birth Defects Monitoring Program. The cross-linked index is an expansion of the ICD-9-CM Congenital Anomalies category 740-759.9 and includes multiple disease descriptions, synonyms, and other descriptive terms that are used to describe birth defects.
- Classification systems provide a framework for coding but often do not provide compatible definitions of diagnoses. Clinical case definitions and case definitions used for public health surveillance are not always the same. Surveillance systems must specify how clinical documentation should be used to determine the appropriate disease code. See Chapter 3 on Case Definition for a discussion of ways to determine how birth defects should be coded.
- ➤ Diagnostic categories are not consistent in the amount of detail they provide, nor are they always clear. For instance, the chromosomal anomaly category (758) is very general. The musculoskeletal system (754-756) is not well-defined. Additionally, all birth defects are not identified with an explicit code, so there can be questions about how to code a particular defect or whether it should be coded at all.
- A single ICD-9-CM code may be used to describe several different defects. This may make it difficult to use the code to recover specific information. For example, codes such as those listed below present challenges because of the potential heterogeneity of the defects included under a given code:
 - 742.2 Reduction deformities of brain (includes holoprosencephaly and absent corpus callosum)
 - 747.21 Anomalies of aortic arch (includes overriding aorta and double aortic arch)
 - 753.0 Renal agenesis and dysgenesis (includes absent kidney and hypoplasia of kidney)
 - 756.0 Anomalies of skull and face bones (includes hypertelorism and craniosynostosis)
 - 756.79 Other congenital anomalies of abdominal wall (includes gastroschisis and omphalocele)
- ➤ ICD-9-CM codes do not reflect the status of the diagnosis. For example, a condition may be possible or probable. This is problematic when birth defects are reported to the surveillance system in coded format, or when programs use the hospital disease index in case finding.
- How information is coded in an administrative database (e.g., hospital disease index, hospital discharge data, Medicaid data) is determined by the methods used to assign codes and by the objectives of those who maintain the database. In other words, code use is defined by the "business operations" of the facility or organization doing the coding. For example, the ICD-9-CM classification system is used primarily in hospitals and other care settings to comply with federal financial justification for payment. Coding decisions made by someone with that goal in mind could be different

from those made by someone coding for a surveillance system.

➤ Professional disease coding training and courses for ICD-9-CM are beneficial in providing a good foundation for training staff regardless of the surveillance approach being used (i.e., active or passive case ascertainment). Information on such courses is available from the American Health Information Management Association (http://www.ahima.org).

5.4 Guidelines for Effective Coding

As noted earlier, the primary goal in coding information is to provide accurate, consistent, and concise representation of that information. Coded diagnostic information is easier to analyze, compare, retrieve, and store. All of these attributes promote the use and dissemination of information between systems. The use of computer technology and the development of particularly large databases have accelerated the demand for coded information. The standardization of information that is translated into a code or discrete data element is one of the objectives of the Health Insurance Portability and Accountability Act of 1996 (HIPAA). For a thorough discussion of birth defects coding, see Rasmussen and Moore (2001).

Programs should:

- ➤ **Develop** well-defined surveillance case definitions. This includes identifying the characteristics of eligibility (e.g., demographics, pregnancy outcome, gestational age), and specific birth defects or diagnosis. These issues are discussed in Chapter 3 on Case Definition.
- ➤ Understand that the disease classification system and associated coding guidelines are developed to standardize results and assist in decision-making. The coding rules for ICD-9-CM as used by hospitals are established at the federal level through a set of guidelines administered, maintained, and updated by NCHS. To comply with these federal standards, a hospital coder may be required to use codes that differ from those used by a surveillance system coder. The 6-digit CDC code is supported by a body of guidelines and procedures that specifically address issues in assigning codes to birth defects. Coding rules for the 6-digit CDC code are detailed and have many exceptions. For example, when using the 6-digit CDC code, there may be exceptions in the laterality rule (i.e., does not apply to all diagnoses).
- ➤ Adapt surveillance procedures and the database to disease code changes as they occur. ICD-9-CM codes and code definitions are subject to rule changes, additions, deletions, and edits. ICD-9-CM changes are usually timed to coincide with the beginning of the federal fiscal year. It is essential for programs that use administrative databases to be aware of these code changes.
- ➤ Track disease code changes. Consider adding a date field to each disease code listed in the database. Disease codes are added, deleted, or edited by the authoritative agency, usually on an annual basis. Any code assignment change may affect statistical analysis or other evaluation activities. Tracking disease code changes will be an essential task when ICD-10-CM replaces ICD-9-CM.
- Assign a disease code to each diagnosis that is reportable to the program. This facilitates building a database of eligible disease codes (conditions), which can be incorporated into abstracting software (e.g., drop-down windows) and used to develop queries and generate lists.
- ➤ **Identify** the disease classification system that is to be used. Some programs may use more than one disease classification system.

Examples:

• An active case ascertainment system might only use the 6-digit CDC code.

- A passive case ascertainment system might only use the ICD-9-CM classification system.
- A passive case ascertainment system might use the 6-digit CDC code if the program receives case reports in a descriptive or literal format and if surveillance staff assign codes.
- A passive case ascertainment system might use ICD-9-CM for case reports that are submitted to the program, but might use the 6-digit CDC code when staff actively review medical charts or for special projects.
- ➤ Promote the use of the 6-digit CDC code where possible. Because the CDC code conveys greater detail, surveillance systems should ideally incorporate this coding system into regular program operations. This may be easier for active ascertainment systems, as passive case ascertainment systems are often limited to the standard classification system in use at hospitals (i.e., ICD-9-CM). However, in order to promote consistency, accuracy, completeness, and comparability across birth defects programs, passive case ascertainment should use the 6-digit CDC code whenever possible.
- ➤ Use the NBDPN Abstractor's Instructions cited in Chapter 3 on Case Definition. This tool should be used as a reference for the birth defects that are central to the NBDPN. It describes the diagnosis and identifies the appropriate disease code.
- ➤ Use technical reference materials. For example, *The International System for Human Cytogenetic Nomanclature (ISCN)* is the definitive guide to understanding the classification system used in cytogenetics (Mitelman, 1995). The text provides information on definitions, on how to read and understand karyotypes, and on other technologies used in laboratory analysis. Surveillance systems can use the *ISCN* as a tool to assist in assigning a disease code to a case with a chromosomal anomaly.
- ➤ Use clinicians for advice on understanding medical conditions and for providing guidance on assignment of disease codes.
- ➤ Develop coding procedures for abstractors, especially as relates to standardized methods for translating medical information into a disease code. Document decision items that result from coding discussions through the use of a decision log or similar record-keeping system. Surveillance systems that are research based may require a different set of procedures than a surveillance system that is focused on providing services. The NBDPN Abstractor's Instructions cited in Chapter 3 on Case Definition provide a good foundation.

5.5 Coded Data Quality Issues

Many factors can affect the quality of coded data. As mentioned earlier, any disease coding system has limitations. Additionally, the translation of a medical diagnosis into a disease code requires interpretation and judgment. Programs can improve the quality of coded data by considering the following recommendations.

Programs should:

- ➤ **Promote** coding to the highest degree of accuracy, completeness, and consistency as required by the surveillance system and as recommended by the NBDPN.
- ➤ **Develop** methods to identify situations that may result in inconclusive or incomplete diagnoses. This is particularly important for programs that work with or receive diagnosis information in coded format. Programs can use length-of-stay patterns, type of diagnosis, and type of data source (e.g., prenatal diagnosis center) to determine whether follow-up is necessary. For example, a chromosomal anomaly diagnosed during the newborn period may be a 'suspect' condition at discharge, pending receipt of laboratory results.
- ➤ Code all individual defects associated with a chromosomal anomaly, syndrome, or association, unless a coding rule or the NBDPN Abstractor's Instructions cited in Chapter 3 on Case Definition specify otherwise (see next recommendation). Code the major chromosomal anomaly or syndrome as well. Some of the most frequently diagnosed syndromes are listed in the category 759.8 in the 6-digit CDC code (see Appendix 5.2).
 - Chromosomal anomalies should be coded to the highest degree of detail that is provided by the karyotype.
 - Birth defects that are components of syndromes identified by 759.8x should be coded separately.
- ➤ Identify those birth defects that are exceptions to the 'code all defects' rule outlined above. For some diagnoses, all birth defects related to the condition may not need to be coded. Refer to the NBDPN Abstractor's Instructions cited in Chapter 3 on Case Definition for a listing and description of these conditions. Develop methods to query the database to find potentially "extra" disease codes. This often occurs with passive case ascertainment using multiple data sources. Some sources may report the major birth defect, while others may report each defect within the major diagnosis.
- ➤ Code at the most specific level possible. For example, if the specific heart defect is known, it is essential to list the specific defect rather than a more general description such as 'congenital heart disease'. Passive case ascertainment systems may find it useful to develop data quality audits to identify diagnoses that frequently are assigned general or non-specific codes and that may merit follow-up.
- ➤ **Develop** computer edit checks to identify problems with code use. For example, some conditions should be combined under a single code. These include spina bifida and hydrocephalus, imperforate anus and anal fistula, esophageal atresia and tracheoesophageal fistula, tetralogy of fallot, and cleft lip and palate. Edit checks can also be developed for gender-specific conditions and for conditions that may also be acquired (e.g., hydrocephalus, skeletal deformations). Edit checks can further be used to identify codes for defects that should not be counted due to gestational age, birth weight, or other established eligibility criteria.

- **Develop** methods for identifying general or non-specific codes, miscodes, inappropriate or redundant codes, or unusual combinations of coded data in a case abstract or case record.
- Evaluate the accuracy and consistency of code assignment. Conduct evaluations to determine the level of agreement in code assignment among program staff, as well as between staff and acute care coders in hospitals. This is particularly effective in identifying differences that result due to federal ICD-9-CM coding guidelines. Identify problem areas and implement quality control procedures as necessary.
- **Develop** coding procedures documentation especially regarding decision items, discussion points, or code assignments. Identify implementation dates.

5.6 Tips and Hints

- Coded data can be used to enhance surveillance capability, as they are easily manipulated and queried in a database. For example:
 - Birth defects case records that have multiple disease codes can be identified and investigated further to determine whether an underlying condition or syndrome is present.
 - Birth defects codes that are included or excluded due to specific criteria can be identified and flagged.
- Administrative databases, especially hospital discharge data, use the ICD-9-CM coding system. Discharge data can be used for specific screening purposes. For example:
 - Maternal pregnancy disease codes may identify potential birth defects cases, especially if the pregnancy results in a fetal demise.
 - Possible cases of birth defects can be queried using disease codes for prematurity, low birth weight, stillbirths, etc.
- > Some programs may find it helpful to retain the complete descriptive text of the birth defect. As previously stated, disease coding systems have limitations. While birth defects are translated to the most accurate disease code, the code may not be precise enough in describing the birth defect.
- ➤ Patterns of disease code assignment for particular birth defects may vary between hospital disease coders. During case finding and abstracting and when reviewing medical records, it is helpful to be observant of coding patterns and inclinations. In many instances, disease codes are listed in the medical records, which helps with these informal assessments.

5.7 References

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Appendix 5.1 Texas Disease Index

The Texas Birth Defects Monitoring Division (TBDMD) created the Texas Disease Index to be used in conjunction with the six-digit codes for reportable birth defects developed by the National Center for Birth Defects and Developmental Disabilities of the Centers for Disease Control and Prevention (CDC). The six-digit birth defect codes, commonly called the BPA code, were developed based on the British Pediatric Association (BPA) Classification of Diseases (1979) and the World Health Organization's International Classification of Diseases, 9th Revision, Clinical Modification (ICD-9-CM) (1979).

The Texas Disease Index was developed for use by the TBDMD, which utilizes active case ascertainment. In addition to being useful to other surveillance programs that carry out active case ascertainment, it is also a valuable resource for systems that have passive case ascertainment based on reporting by standard ICD-9-CM codes.

It should be noted that the TBDMD made some modifications to the BPA code list. Therefore the Texas Disease Index may deviate slightly from the six-digit CDC code list used by other active case ascertainment surveillance programs, which is included as an appendix to these guidelines by reference to the website. Most of the modifications relate to birth defects that were not listed explicitly in the original BPA codes. These additional birth defects have been reviewed by various TBDMD staff, including two clinical geneticists, and appropriate BPA codes have been assigned to them.

Note that for ease of use a diagnosis may be listed in more than one format in this index. For example, 'absent eye' may be found under 'absent, eye' or 'eye, absent.

The TBDMD revises this index periodically, indicating the revision date on each page. New revisions will be made available through the surveillance guidelines and standards webpage.

This document may be viewed or downloaded at the NBDPN website at: http://www.nbdpn.org/bdsurveillance.html

References

British Paediatric Association (BPA). *Classification of Diseases*. London, England: British Paediatric Association; 1979.

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1

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Aarskog syndrome - 759.800
Abdominal
        cvst NOS - 759.990
        mass NOS - 759.990
Abdominal wall
        benign neoplasm - # 216.500
        other and unspecified anomalies - 756.790
Abduction
        foot - L 754.690
       hip - x
Aberrant
        innominate artery - L 747.640
        subclavian artery - L 747.640
Ablepharon - L * 743.630
Absent - see also agenesis, atresia
        adrenal gland - L 759.100
        alimentary tract, NOS (complete or partial) - 751.800
        anus
                with fistula - 751.230
                without fistula - 751.240
        aorta - 747.200
        aortic valve - 746.480
        appendix - 751.200
        arm - L 755.200
        auditory canal (without hypoplastic pinna) - L 744.000
        auricle - L 744.010
        bladder - 753.800
        brain - 740.000
        breast
                nipple absent - L 757.600
                nipple present - L 757.610
        broad ligament - L 752.100
        bronchus - L 748.350
        carotid artery - L 747.640
        cervix (genital) - 752.400
        clitoris - * 752.450
        colon - 751.200
        diaphragm - L 756.600
        digestive system, NOS (complete or partial) - 751.800
        digit, NOS - L 755.440
        duodenum - 751.100
        ear - L 744.010
        ear canal (without hypoplastic pinna) - L 744.000
        external genitalia
                female - * 752.440
                male - 752.880
        eye - L 743.000
        eyebrow - L 744.880
        eyelash - L * 743.630
        eyelid - L * 743.630
        face - L 744.880
        fallopian tube - L 752.100
        femur (total or partial)
                only - L 755.320
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with absent tibia and fibula (total or partial)- L 755.310
        with absent tibia, fibula, and foot - L 755.300
fibula
        only (total or partial) - L 755.366
        with absent femur (total or partial) and tibia - L 755.310
        with absent femur (total or partial), tibia, and foot - L 755.300
        with absent tibia - L 755.320
        with absent tibia and foot - L 755.330
finger
        fifth (with or without fourth) - L 755.270
        first (thumb) - L 755.260
        first (thumb) and absent radius (total or partial) - L 755.260
        NOS - L 755.240
        third (with or without second, fourth) - L 755.250
        with absent forearm long bone - L 755.265
fontanelle - # 754.040
foot
        only - L 755.340
        with absent femur (total or partial), tibia, and fibula - L 755.300
        with absent lower leg - L 755.330
        with absent tibia and fibula (total or partial) - L 755.330
forearm
        long bone with absent fingers - L 755.265
        only - L 755.220
        with absent hand - L 755.230
        with absent upper arm - L 755.210
foreskin - 752.860
genitalia (sex unknown) - * 752.790
hand
        only - L 755.240
        with absent forearm - L 755.230
        with absent humerus (total or partial), radius, and ulna - L 755.200
        with absent radius and ulna (total or partial) - L 755.230
head - 740.080
humerus (total or partial)
        only - L 755.220
        with absent radius, and ulna - L 755.210
        with absent radius, ulna, and hand - L 755.200
ileum - 751.120
intestine
        large - 751.200
        small - 751.190
        small, with fistula - 751.195
iris - L 743.420
jejunum - 751.110
kidney
        bilateral - 753.000
        NOS - 753.009
        unilateral - L 753.010
lacrimal apparatus - L 743.640
leg - L 755.300
lens - L 743.300
limb. NOS - L 755.400
liver, total or partial - 751.600
long bone leg with absent toe - L 755.360
lower leg
        only - L 755.320
```

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with absent foot - L 755.330
        with absent thigh - L 755.310
luna - L 748.500
meatus (external auditory, ear) - L 744.000
mitral valve - 746.505
muscle - L 756.810
nail - L 757.500
nares - 748.100
nasal septum - # 748.180
neck - # 744.900
nipple
        only - L 757.630
        with absent breast - L 757.600
nose - 748.100
olfactory nerve - 742.270
ovary - L 752.000
palate
        hard - 749.030
        NOS - 749.090
        soft - 749.070
pancreas - 751.700
patella - L 755.647
penis - 752.850
phalange (isolated)
        finger - L 755.240
        toe - L 755.340
pinna (ear) - L 744.010
pulmonary arteriovenous - L 747.340
pulmonary artery - L 747.300
pulmonary valve - 746.000
punctum lacrimale - L 743.640
radius
        only (total or partial) - L 755.260
        with absent humerus (total or partial) and ulna - L 755.210
        with absent humerus (total or partial), ulna, and hand - L 755.200
        with absent thumb - L 755.260
        with absent ulna - L 755.220
rectum
        with fistula - 751.210
        without fistula - 751.220
renal artery - L 747.610
respiratory organ NOS - 748.900
rib - L 756.300
right superior vena cava - x
septum between aorta and pulmonary artery - 745.000
septum pellucidum - 742.210
skin - 757.395
spleen - 759.000
sternocleidomastoid muscule - L 754.100
sternum - 756.350
stomach
        with absent GI tract - 750.780
        with rest of GI tract intact - 750.700
superior vena cava, right - x
tarsal bones - L 755.340
tendon - L 756.820
testicle - L 752.800
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thigh and lower leg - L 755.310
        thymus - * 759.240
        tibia
                only (total or partial) - L 755.365
                with absent femur (total or partial) and fibula (total or partial)- L 755.310
                with absent femur (total or partial), fibula, and foot - L 755.300
                with absent fibula - L 755.320
                with absent fibula (total or partial) and foot - L 755.330
                with absent first toe (with or without second toe) - L 755.365
        toe
                fifth (with or without fourth) - L 755.366
                first toe (with or without second toe) - L 755.365
                first toe (with or without second toe) and tibia (total or partial) - L 755.365
                NOS - L 755.340
                third (with or without second, fourth)- L 755.350
                with absent long bone leg - L 755.360
        tongue - 750.100
        ulna
                only (total or partial) - L 755.270
                with absent humerus (total or partial) and radius - L 755.210
                with absent humerus (total or partial), radius, and hand - L 755.200
                with absent radius - L 755.220
        upper arm
                only - L 755.220
                with absent forearm - L 755.210
        ureter - L 753.400
        urethra - 753.800
        uterus - 752.300
        uvula - 749.080
        vagina (complete or partial) - 752.410
        vena cava (except left superior) - 747.480
        vulva - * 752.440
Acardiac twins - 759.480
Accessory - see also extra
        adrenal gland - L 759.120
        auricle - L # 744.100
        carpal bone - L 755.525
        breast (with accessory nipple) - L 757.620
        digit - see polydactyly
        finger - see polydactyly
        kidney - L 753.300
        lung lobe - L 748.620
        nipple
                only - L # 757.650
                with accessory breast - L 757.620
        nose - 748.110
        ovary - L 752.020
        pancreas - 751.710
        spleen - 759.040
        toe - see polydactyly
        ureter - L 753.410
Achalasia of cardia - 750.720
Achilles tendon, short - L 754.720
Achondrogenesis
        type I - 756.480
        type II - 756.480
Achondroplastic dwarfism - 756.430
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Acne, neonatal - x
Acrania - 740.010
Acrocallosal syndrome - 759.890
Acrocephalosyndactyly
        NOS - 756.050
        other specified - 756.057
        type I - 756.055
        type II - 756.055
        type III - 756.056
Acrocephaly - 754.080
Acrodactylia
        finger - L # 755.500
        toe - L # 755.600
Acyanotic congenital heart disease - 746.920
Adams-Oliver syndrome - 759.840
Adduction foot - L 754.590
Adductus
        metatarsus - L # 754.520
Adhesion of omentum and peritoneum - 751.420
Adrenal gland
        absent - L 759.100
        accessory - L 759.120
        dysgenesis - L 759.180
        ectopic - L 759.130
        enlarged - L 759.180
        fused - L 759.180
        hyperplasia, congenital
                classical (salt) water - # 255.200
                classical (simple virilizer) - # 255.210
                NOS - # 255.290
                other than 21-OHP deficiency - # 255.240
        hypoplasia - L 759.110
        other specified anomalies - L 759.180
        unspecified anomalies - L 759.190
Adrenogenital syndrome - # 255.290
Aganglionosis of intestine
        beyond the rectum - 751.310
        involving no more than the anal sphincter and the rectum - 751.320
        total - 751.300
Agenesis - see also absent
        bile duct - 751.650
        cervix (genital) - 752.400
        gallbladder - 751.630
        hepatic duct - 751.650
        kidney
                bilateral - 753.000
                NOS - 753.009
                unilateral - L 753.010
        liver, total or partial - 751.600
        lung - L 748.500
        nose - 748.100
        ovary - L 752.000
        pancreas - 751.700
        uterus - 752.300
        vagina (complete or partial) - 752.410
        vertebrae
                cervical - 756.146
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lumbar - 756.166
               sacral - 756.170
               thoracic - 756.156
Aglossia - 750.100
Agnathia - * 524.000
Agnathia formation complex - 759.800
Agyria - 742.240
Aicardi syndrome - 759.890
Alae nasae hypoplasia - # 748.180
Alagille syndrome - 759.870
Albers-Schonberg syndrome - 756.540
Albinism - # 270.200
Albright-McCune-Sternberg syndrome - 756.510
Alimentary tract
        absent (complete or partial) - 751.800
       duplication - 751.810
       ectopic - 751.820
       obstruction, NOS - 752.900
       other specified anomalies - 751.880
       unspecified anomalies - 751.900
Almond shaped eye - L # 743.800
Alopecia - 757.400
Alport syndrome - 759.870
Ambiguous genitalia - * 752.790
Amelia
        arm - L 755.200
       leg - L 755.300
       limb, NOS - L 755.400
Amniotic
       bands - # 658.800
       cyst - # 658.800
Amputation, NOS
       arm - L 755.285
       leg - L 755.385
       limb, NOS - L 755.420
Amsterdam dwarf - 759.820
Amyelia - 742.500
Amyoplasia congenita - 756.840
Amyotrophia congenital - 756.840
Anasarca - # 778.000
Androgen insensitivity syndrome - 257.800
Anencephaly - 740.020
       other - 740.080
Aneurysm
       aorta - 747.270
       arteriovenous (brain) - L 747.800
        atrial septum - x
       pulmonary artery - 747.330
       sinus of Valsalva - 747.240
       tricuspid valve - 746.100
Angelman syndrome - 759.890
Angulation of tibia - L * 755.630
Aniridia - L 743.420
Anisocoria - L 743.440
Ankle
       anomalies - L 755.620
       other specified deformities - L 754.780
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Ankyloblepharon - L * 743.630
Ankyloglossia - # 750.000
Annular pancreas - 751.720
Anomalous portal vein termination - 747.440
Anomalous pulmonary venous return
        partial - 747.430
        total - 747.420
        total/partial not specified - 747.480
Anonychia - L 757.500
Anophthlmos - L 743.000
Anotia - L 744.010
Anovaginal fistula - 752.420
Anterior
        frenulum (tongue, lingual) - # 750.000
        segment of eye
                other specified colobomas - L 743.480
                other specified anomalies - L 743.480
                unspecified anomalies - L 743.490
        urethral valve - 753.620
Anteversion of femur - L 755.650
Antimongolian syndrome - 758.300
Antimongoloid slant to eyes - L # 743.800
Anus/anal
        absent
                with fistula - 751.230
                without fistula - 751.240
        atresia
                with fistula - 751.230
                without fistula - 751.240
        benign neoplasm - # 216.500
        displaced - 751.530
        duplication - 751.500
        dysgenesis with fistula - 751.230
        dysgenesis without fistula - 751.240
        ectopic - 751.530
        fissure - x
        fistula - 751.540
        imperforate
                with fistula - 751.230
                without fistula - 751.240
        stenosis
                with fistula - 751.230
                without fistula - 751.240
Aorta/Aortic
        artery
                absent - 747.200
                absent septum between pulmonary artery and - 745.000
                aneurysm - 747.270
                atresia - 747.200
                coarctation
                        distal - 747.110
                        juxtaductal - 747.190
                        postductal - 747.110
                        preductal - 747.100
                        proximal - 747.100
                        unspecified - 747.190
                collateral vessel involving - 747.280
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dextroposition - 747.260
                dilatation - 747.270
                double arch - 747.250
                enlarged - 747.270
                hypoplasia - 747.210
                interrupted arch - 747.215
                large - 747.270
                malaligned - 747.260
                narrow - 747.210
                other specified anomalies - 747.280
                overriding - 747.260
                pseudocoarctation - 747.280
                right arch - 747.230
                small - 747.210
                supra-aortic stenosis - 747.220
                supravalvular - 747.220
                unspecified - 747.290
        NOS
                septal defect - 745.010
                stenosis - 746.300
                subvalvular stenosis - 746.300
        valve
                abnormal - 746.490
                absent - 746.480
                atresia - 746.480
                bicuspid - * 746.400
                dysmorphic - 746.480
                dysplastic - 746.480
                hypoplastic - 746.480
                incompetence - * 746.400
                insufficiency - * 746.400
                other specified - 746.480
                quadricuspid - 746.480
                regurgitation - * 746.400
                small - 746.300
                stenosis - 746.300
                thickened - 746.480
                unspecified - 746.490
Aortic annulus - see aortic valve
Aortopulmonary window - 745.010
Apert syndrome - 756.055
Aphakia - L 743.300
Aplasia - see also absent, agenesis
        cutis
                not involving scalp - 757.395
                scalp - 757.800
        eye - L 743.100
        penis - 752.850
        red cell - # 284.000
        scrotum - L * 752.810
        testicle - L * 752.810
Appendix
        absent - 751.200
        atresia - 751.200
        duplication - 751.500
        stenosis - 751.200
        testicle - L 752.870
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transposition - 751.510
Aqueductal stenosis (without spina bifida) - 742.300
Aqueduct of Sylvius anomalies without spina bifida - 742.300
Arachnodactyly
        finger - L # 755.500
        toe - L # 755.600
Arachnoid cyst - x
Arm
        absent - L 755.200
        amelia - L 755.200
        amputation, NOS - L 755.285
        benign neoplasm - L # 216.600
        hyperextensibility - L 755.580
        hypomelia - L 755.585
        hypoplasia - L 755.585
        intercalary reduction defect - L 755.210
        long - x
        longitudinal reduction defect
                NOS - L 755.265
                postaxial - L 755.270
                preaxial - L 755.260
        other anomalies (whole) - L 755.560
        other specified anomalies - L 755.580
        other specified reduction defect - L 755.280
        phocomelia - L 755.210
        positional deformity - L 755.580
        short - L 755.580
        transverse reduction defect, NOS - L 755.285
        unspecified anomalies - L 755.590
        unspecified reduction defect - L 755.290
Arnold-Chiari malformation
        with spina bifida - 741.010
        without spina bifida - 742.480
Arrhinencephaly - 742.270
Arrhythmias, cardiac, NOS - 427.900
Arteriovenous malformation
        brain - L 747.800
        peripheral - L 747.620
Arthrogryposis multiplex congenita - L 755.800
Ascites, congenital - # 778.000
Asphyxiating thoracic dystrophy - 756.400
Asplenia - * 759.000
Association - see syndrome
Astragaloscaphoid synostosis - L 755.620
Asymmetry
        brain - x
        calvarium - 754.055
        chest - 754.820
        crying facies - L 351.000
        ears - x
        eyes - x
        face - 754.000
        gluteal cleft - x
        head - 754.055
       jaw - * 756.080
        mouth - L 744.880
        nipples - # 757.680
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nose - # 748.180
        skull - 754.055
Atelomyelia - 742.510
Atresia
        anus
                with fistula - 751.230
                without fistula - 751.240
        aorta - 747.200
        aortic valve - 746.480
        appendix - 751.200
        bile duct - 751.650
        biliary - 751.650
        bladder neck - 753.610
                other and unspecified - 753.690
        cervix (genital) - 752.400
        choanal - L 748.000
        colon - 751.200
        duodenum - 751.100
        esophageal
                without tracheoesophageal fistula - 750.300
                with tracheoesophageal fistula - 750.310
        hepatic duct - 751.650
        ileum - 751.120
        intestine
                large - 751.200
                small - 751.190
                small, with fistula - 751.195
        jejunum - 751.110
        lung - L 748.500
        meatus (urethral, urinary) - 753.630
        mitral valve - 746.505
        nares - L 748.000
        piriform aperature - L 748.000
        pulmonary
                artery
                        without septal defect - L 747.300
                        with septal defect - L 747.310
                NOS (heart) - 746.995
                valve - 746.000
                vein - 747.480
        pyloric - 751.100
        rectum
                with fistula - 751.210
                without fistula - 751.220
        trachea - 748.330
        tricuspid valve - 746.100
        ureter - L 753.210
        urethra
                anterior - 753.620
                other and unspecified - 753.690
        urinary meatus - 753.630
        vagina (complete or partial) - 752.410
        vas deferens - L 752.830
Atrioventricular canal
        common - * 745.630
        common, with VSD - * 745.620
        complete - * 745.630
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complete, with VSD - * 745.620
Atriventricular septal defect - see atriventricular canal
Atrioventricular valve
        left - see mitral valve
        right - see tricuspid valve
        single - 746.900
                insufficiency - 746.900
                regurgitation - 746.900
Atrium/atrial
        common - 745.610
        dilatation - x
        enlarged - x
        hypoplastic - 746.887
        inversion - 746.880
        other defects - 746.887
        septal defect
                aneurysm - x
                fenestrated - 745.510
                fossa ovalis - 745.510
                NOS - * 745.590
                ostium primum - * 745.600
                ostium secundum - 745.510
                other specified - 745.580
                primum - * 745.600
                secundum - 745.510
                vs PFO - * 745.590
        single - 745.610
Atrophy
        cerebellar - 742.230
        cerebral - 742.480
        cortical (brain) - 742.480
        muscle (specified muscle) - L 756.880
        optic nerve - L 743.520
        testicle - L * 752.810
        umbilicus - # 759.900
        vermian - 742.230
Auditory canal
        absent - L 744.000
        benign neoplasm - L # 216.200
        small - L 744.000
        stenosis - L 744.000
Auditory meatal stenosis - L 744.000
Auricle - see pinna
Auricular
        pit (ear) - L # 744.410
        septal defect (heart) - * 745.590
Autosome (chromosome)
        deletion - see deletion
        marker - 758.580
        mosaic - see mosaic
        other specified anomalies - 758.580
        translocation - see translocation
        trisomy - see trisomy
        unspecified anomalies - 758.590
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Balantic hypospadias - 752.605
Balantic hypospadias with chordee - 752.625
Baller-Gerold syndrome - 759.840
Band
        amniotic - # 658.800
        heart, anomalous - 746,910
        intestine - 751.420
        Ladd's - 751.420
        omentum - 751.420
        peritoneum - 751.420
Barrel chest - 754.820
Bart syndrome - 757.330
Basilar craniosynostosis - 756.030
Bat ear - L # 744.220
Bathocephaly - * 756.080
Beaded hair - 757.410
Beals syndrome - 759.860
Beckwith syndrome - 759.870
Beckwith-Wiedemann syndrome - 759.870
Beemer Langer syndrome - 759.860
Bell shaped chest - 754.820
Bell's palsy - L # 351.000
Benign external hydrocephaly - x
Bent nose - # 754.020
Bicornate uterus - L 752.380
Bicuspid
        aortic valve - 746.400
        pulmonary valve - 746.080
       tricuspid valve - 746.100
Bifid - see also cleft, accessory
       nose - 748.120
        rib - L 756.310
        scrotum - 752.820
        sternum - 756.380
        thumb - L 755.010
        uvula - 749.080
        vertebrae
                cervical - 756.140
               lumbar - 756.160
               NOS - 756.180
               sacral - 756.170
               thoracic - 756.150
        xyphoid process - 756.380
Bilateral superior vena cava - 747.410
Bile duct
        agenesis - 751.650
        atresia - 751.650
        other anomalies - 761.670
Biliary
        atresia - 751.650
        dysgenesis - 751.670
        obstruction - x
Biliary tract anomalies, NOS - 751.680
Bilirubin excretion disorders - # 277.400
Bilobar right lung - 748.625
Biparietal narrowing - * 756.080
Birthmark, NOS - # 757.385
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Bitemporal narrowing - * 756.080
Bladder
        absent - 753.800
        cystocele - 753.820
        diverticulum - 753.820
        ectopic - 753.810
        enlarged - x
        exstrophy - 753.500
        extroversion - 753.500
        hernia - 753.820
        hypertrophy - x
        hypoplasia - 753.880
        hypoplastic - 753.880
        neck
               atresia - 753.610
               other and unspecified atresia and stenosis - 73.690
               stenosis - 753.610
        neurogenic - x
        other specified anomalies - 753.880
        outlet obstruction - 753.690
        prolase (mucosa) - 753.830
        small - x
        thickened - x
        trabeculated - x
        unspecified anomalies - 753.920
Blepharophimosis - L 743.635
Blepharophimosis syndrome - 759.800
Blepharoptosis - L 743.600
Block, heart - 746.870
Bloom syndrome - 759.890
Blue
        baby - 746.930
        Mongolian spot - x
        nevus - see skin-benign neoplasm
        sclera - L * 743.450
Blueberry muffin spots - x
BOR syndrome - 759.800
Body stalk anomaly - 756.790
Bone
        unspecified anomalies - 756.920
Bonneville-Ullrich syndrome, NOS - 758.690
Bourneville's disease - 759.500
Bowed/bowing
        femur - L 754.400
        legs, NOS - 754.420
        lip - L 744.880
        lower leg - L 754.410
        fibula - L 754.410
        tibia - L 754.410
        ulna without Madelung deformity - L 755.530
Box shaped head - 754.080
Brachial plexus palsy - L # 767.600
Brachiocephalic trunk, common - L 747.640
Brachycephaly - 754.080
Brachydactyly
        finger - L # 755.500
        toe - L # 755.600
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Bradycardia - x
Brain
        absent - 740.000
        asymmetry - x
        atrophy - 742.480
        enlarged - * 742.400
        other specified anomalies - 742.480
        small - 742.486
        unspecified - 742.900
Brainstem
        anomalies - 742.480
        hypoplastic - 742.280
        reduction defect - 742.280
        small - 742.280
Branch pulmonary artery stenosis - L * 747.325
Branchial arch syndrome - 759.800
Branchial cleft
        cvst - L 744.400
        fistula - L 744.400
        other anomalies - L 744.480
        pit - L 744.400
        remnant - L 744.400
        sinus - L 744.400
Breast
        absent
                nipple absent - L 757.600
               nipple present - L 757.610
        accessory (with accessory nipple) - L 757.620
        benign neoplasm - # 216.500
        ectopic (with nipple) - L 757.620
        hypertrophy - x
        hypoplastic (with hypoplastic nipple) - L 757.610
        other specified anomalies - # 757.680
        small - x
Broad
        face - 744.910
        hand - L 755.510
        neck - # 744.500
Broad ligament
        absent - L 752.100
        other and unspecified anomalies - L 752.190
Bronchiectasis - L 748.610
Bronchoesophageal fistula - 750.330
Bronchogenic cyst - L 748.350
Bronchomalacia - x
Bronchopulmonary dysplasia - x
Bronchopulmonary fistula - L 748.350
Bronchus
        absent - L 748.350
        other anomalies - L 748.350
        other specified anomalies - L 748.380
        stenosis - L 748.340
        unspecified anomalies - 748.390
Brown syndrome - # 378.000
Brushfield spots - L # 743.800
Bulging eye - L # 743.800
Bullosa
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epidermolysis - 757.330
       ichthyosis - 757.115
Bullous type ichthyosis congenita - 757.115
Buphthalmos - L 743.200
Buried penis - 752.860
Butterfly vertebra
       cervical - 756.140
       lumbar - 756.160
       NOS - 756.180
       sacral - 756.170
       thoracic - 756.150
-C-
Café au lait spots - # 757.390
Caffey syndrome - 756.530
Calcaneovalgus - L 754.600
Calcaneovarus - L 754.510
Calvarium - see aso skull
       absent - 740.020
       asymmetry - 754.055
Camptodactyly
       finger - L # 755.500
       toe - L # 755.600
Camptomelic dysplasia - 756.480
Camurati-Engelmann syndrome - 756.550
Canal of Nuck cyst - 752.470
Cardiomegaly - * 746.860
Cardiomyopathy - * 746.860
Cardiomyopathy, hypertrophic - * 746.860
Cardiospasm - 750.720
Cardio-splenic syndrome - 759.890
Cardiovascular system, other specified anomalies - L 747.880
Carotid artery
       absent - L 747.640
Carpal bone
       accessory - L 755.525
Carpenter syndrome - 759.840
Carp shaped mouth - L 744.880
Cartilage (ear)
       absent - L * 744.230
       decreased - L * 744.230
       unspecified anomalies - 756.930
Cat eye syndrome - 758.580
Cataract
       anterior polar - L 743.325
       NOS - L 743.320
       other specified - L 743.326
Cauda equina anomalies, other - 742.530
Caudal dysplasia - 759.840
Caudal regression syndrome - 759.840
Cauliflower ear - L * 744.230
Cavum septum pellucidum - x
Cebocephaly - 759.800
Cecum
       duplication - 751.500
       malrotation - 751.400
```

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Central nervous system (CNS) hemorrhage - x
Cephalohematoma - x
Cephalopagus conjoined twins - 759.410
Cerebellar atrophy - 742.230
Cerebellum anomalies - 742.230
Cerebral/cerebrum
       atrophy - 742.480
       cortical dysplasia - 742.480
       cyst - 742.420
       lipidoses - # 330.100
       reduction deformities - 742.200
Cerebral vessels, other anomalies - L 747.810
Cerebro-oculo-facial-skeletal syndrome - 759.890
Cervical rib - L # 756.200
Cervix (genital)
       absent - 752.400
       agenesis - 752.400
       atresia - 752.400
       doubling - * 752.480
       other specified anomalies - * 752.480
        unspecified anomalies - 752.490
Chalasia - x
CHARGE association - 759.890
Chediak-Higashi syndrome - 757.300
Cheek
       hypoplastic -L 744.880
       skin tag - L # 744.110
Chest
       asymmetry - 754.820
       barrel - 754.820
       bell shaped - 754.820
       benign neoplasm - # 216.500
       deformed - 754.820
       funnel - 754.810
       narrow - 754.820
       other anomalies - 754.820
       pigeon - 754.800
       shield - 754.825
       small - 754.820
Chin
       cleft - x
       dimple - x
       pointed - * 756.080
       receding - 524.000
       small - 524.000
Choanal
       atresia - L 748.000
       stenosis - L 748.000
Choledochal cyst - 751.660
Chondroectodermal dysplasia - 756.520
Chondrodysplasia - 756.410
       other specified - 756.480
       punctata - 756.575
       with hemangioma - 756.420
Chondrodystrophy
       other specified - 756.480
```

```
unspecified - 756.490
Chordee (penile)
        with hypospadias
                coronal - 752.625
               first degree - 752.625
               glandular - 752.625
               NOS - 752.620
               penile - 752.626
               perineal - 752.627
                scrotal - 752.627
               second degree - 752.626
                third degree - 752.627
        without hypospadias - 752.621
Choroid (eye)
        coloboma - L 743.535
        specified anomalies - L 743.530
Choroid plexus cyst
        bilateral - * 742.485
        multiple - * 742.485
        unilateral - x
Chorioretinitis - # 363.200
Chromosome
        autosome - see autosome
        NOS
               additional, NOS - 758.910
               deletion, NOS - 758.920
               duplication, NOS - 758.930
               mosaicism, NOS - 758.900
        unspecified anomaly - 758.990
        sex - see sex chromosome
Chylothorax - # 457.800
Circulatory system, unspecified anomalies - 747.900
Cisterna magna, enlarged - 742.380
Clavicle anomalies - L 755.550
Claw
        foot - L 755.350
        hand - L 755.250
Cleft
        alveolar ridge/alveolus - 749.100
        branchial - L 744.400
        chin - x
        ear - L * 744.230
        face/facial - L 744.880
        foot - L 755.350
        gingiva - 749.100
        gum - 749.100
        hand - L 755.250
        laryngotracheoesophageal - 748.385
        larynx - 748.385
        lip
               lateral - 744.800
               with any cleft palate - L 749.200
                        central - 749.220
                        midline - 749.220
               without cleft palate - L 749.100
                       central - 749.120
                        midline - 749.120
```

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mandible - * 756.080
        mitral valve - 746.505
        mouth, lateral - 744.800
        nose - 748.120
        palate
                with cleft lip - see cleft lip with any cleft palate
                without cleft lip
                        hard palate (alone) - L 749.000
                                central - 749.020
                                midline - 749.020
                        NOS (hard/soft not specified) - 749.090
                        soft and hard palate - 749.090
                        soft palate (alone) - L 749.040
                                central - 749.060
                                midline - 749.060
                        submucosal
                                hard - 749.020
                                NOS (hard/soft not specified) - 749.090
                                soft - 749.060
        tongue - 750.140
        tricuspid valve - 746.100
        uvula - 749.080
        vertebrae
                cervical - 756.140
                lumbar - 756.160
                NOS - 756.180
                sacral - 756.170
                thoracic - 756.150
Cleidocranial dysostosis - 755.555
Clenched hand or fist - L # 755.500
Clenched toes - L # 755.600
Click, hip - x
Clifford's syndrome - x
Clinodactyly
        finger - L # 755.500
        toe - L # 755.600
Clitoris
        absent - * 752.450
        enlarged - * 752.450
        hypertrophy - * 752.450
        other anomaly - * 752.450
        prominent - * 752.450
        prominent prepuce - x
Clitoromegaly - * 752.450
Cloaca
        exstrophy - 751.550 and 756.790
        persistent - 751.550
Close set eyes - * 756.080
Cloudy cornea - L 743.400
Cloverleaf head shape - 756.000
Club/clubbed
        fingers - L 754.840
        foot, NOS - L 754.730
        hand - L 754.840
        nail - L 757.540
Coarctation of aorta
        distal - 747.110
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juxtaductal - 747.190
        postductal - 747.110
        preductal - 747.100
        proximal - 747.100
        unspecified - 747.190
Cockayne syndrome - 759.820
Coffin-Siris syndrome - 759.800
COFS syndrome - 759.890
Collateral vessel
        involving aorta - 747.280
        involving pulmonary artery (and not aorta) - L 747.380
        not involving aorta or pulmonary artery - L 747.880
Collodian baby - 757.110
Coloboma
        anterior segment
               other - L 743.480
                unspecified - L 743.490
        choroid - L 743.535
        eyelid - L 743.636
        iris - L 743.430
        lens - L 743.340
        NOS - L 743.490
        optic disc/nerve - L 743.520
        retina - L 743.535
Colon
        absent - 751.200
        atresia - 751.200
       hypoplastic - 751.520
        malrotation - 751.400
        short - 751.520
        small - 751.520
        stenosis - 751.200
        transposition - 751.510
Colpocephaly - 742.280
Common
        atrioventricular canal - * 745.630
        atrioventricular canal with VSD - * 745.620
        atrium - 745.610
        brachiocephalic trunk - L 747.640
        ventricle (heart) - 745.300
Complete
        atrioventricular canal - * 745.630
        atrioventricular canal with VSD - * 745.620
        mirror reversal of abdominal organs with normal thoracic organs - 759.330
        mirror reversal of all organs - 759.300
        mirror reversal of thoracic organs with normal abdominal organs - 759.320
Complex - see syndrome
Concealed penis - 752.860
Conduction defects (heart) - 746.880
Cone shaped head - 754.080
Congenital anomaly, NOS - 759.990
Congenital contractural arachnodactyly syndrome - 759.860
Congenital encephalopathy - x
Congenital heart disease
        acyanotic - 746.920
        cyanotic - 746.930
        NOS - 746.990
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Conjoined twins
        cephalopagus - 759.410
        craniopagus (head-joined twins) - 759.410
        dicephalus (two heads) - 759.400
        ischiopagus - 759.480
        other specified - 759,480
        pelvis-joined twins - 759.480
        pygophagus (buttock-joined twins) - 759.440
        thoracopagus (thorax-joined twins) - 759.420
        unspecified - 759.490
        xiphopagus (xiphoid-joined twins) - 759.430
Conjunctivitis - x
Connective tissue
        other specified anomalies - L 756.880
        unspecified anomalies - 756.940
Conradi syndrome - 756.575
Constriction band syndrome - # 658.800
Contracture
        joint (flexion, individual) - L 755.800
        sternocleidomastoid muscule - L 754.100
Cor biloculare - 745.700
Cornea
        cloudy - L 743.400
        enlarged - L 743.220
        leukoma - L 743.400
        opacity - L 743.400
        other specified - L 743,410
Cornelia de Lange syndrome - 759.820
Coronal suture
        closed - L 756.010
        craniosynostosis - L 756.010
        fused - L 756.010
Coronary artery anomalies - 746.885
Coronary sinus anomalies - 746.885
Corpus callosum
        anomalies - 742.210
        cvst - 742.420
Cortex/cortical
        anomalies - 742.200
        atrophy - 742.480
        dysplasia (cerebral) - 742.480
        hyperostosis, infantile - 756.530
Cor triatriatum - 746.820
Cor triloculare biatriatum - 745.300
Costello syndrome - 759.800
Coxa
        valga - L 755.660
        vara - L 755.660
Cranial nerve defects - 742.480
Craniofacial
        abnormality NOS - 756.090
        craniofacial disproportion - 756.090
        dysostosis - 756.040
        other syndromes - 756.046
Craniorachischisis - 740.100
Cranioschisis - 740.020
Craniosynostosis
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basilar - 756.030
        coronal - L 756.010
        lambdoidal - L 756.020
        metopic - 756.006
        NOS - 756.000
        other - 756.030
        sagittal - 756.005
        squamosal - 756.000
Craniotabes - x
Cranium, square - 754.080
Crease
        ear - L 744.280
        infraorbital - L # 743.800
        palm or hand - see palmar crease
Crepitus hip - x
Cri du chat syndrome - 758.310
Cross fused renal ectopia - 753.320
Crossed eyes - # 368.000
Crouzon's disease - 756.040
Cryptophthalmos - L 743.000
Cryptorchidism
        bilateral - * 752.514
        left - L * 752.501
        NOS - * 752.520
        right - L * 752.502
        unilateral - L * 752.500
Cubitus valgus - L 755.540
Curvature of spine (postural), NOS - 754.220
Curved sternum - 754.820
Cutis aplasia
        not involving scalp - 757.395
        scalp - 757.800
Cutis laxa hyperelastica - 757.370
Cutis marmorata - x
Cyanotic congenital heart disease - 746.930
Cyclops - 759.800
Cyst/cystic
        abdominal NOS - 759.990
        adenomatoid malformation lung - L 748.480
        amniotic - # 658.800
        arachnoid - x
        branchial cleft - L 744.400
        bronchogenic - L 748.350
        canal of Nuck - 752.470
        cerebral - 742.420
        choledochal - 751.660
        choroid plexus
                bilateral - * 742.485
               multiple - * 742.485
                unilateral - x
        corpus callosum - 742.420
        dysplasia kidney - L 753.160
        duplication - 751.500
        embryonal (vagina) - # 752.460
        embryonic remnants (male) - L 752.870
        enterogenous - 751.500
        ependymal - 742.420
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epoophoron - L 752.110
        fimbrial - L 752.120
        Gartner's duct - L 752.110
        glioependymal - 742.420
        gum - x
        hydatid of Morgagni - L 752.870
        hygroma - 228.100
        intracranial - 742.420
        kidney (single) - L 753.100
        lacrimal apparatus/duct - L 743.660
        liver - 751.610
        lung
               multiple - L 748.410
               other specified - L 748.480
               single - L 748.400
        mediastinum - 748.810
        mesenteric remnant - L 752.110
        ovarian
               multiple - L 752.085
               single - L 752.080
        pancreatic - 751.740
        parovarian - L 752.120
        periventricular - 742.420
        porencephalic - * 742.410
        posterior fossa - 742.230
        preauricular - L # 744.410
        renal (single) - L 753.100
        skin - # 757.390
        spleen - 759.080
        subependymal - 742.420
        thyroglossal - 759.220
        tonque - x
        urachus - # 753.700
        vagina
               embryonal - # 752.460
               other - 752.470
        ventricular (brain) - * 742.485
        vulva - 752.470
        Wharton duct - x
        Wolffian duct - L 752.870
Cystic fibrosis, no mention of meconium ileus - # 277.000
Cystic fibrosis, with mention of meconium ileus - # 277.010
Cystic kidney NOS - L 753.180
Cystocele bladder - 753.820
Cytomegalovirus (CMV), congenital (in utero infection) - # 771.100
Dacryocystocele - L 743.660
Dacryostenosis - L # 743.650
Dandy-Walker syndrome - * 742.310
Deafness, congenital - L * 744.090
Defect
        Gerbode - 745.420
Deletion (chromosome)
        4 - 758.320
        5 - 758.310
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-D-

```
13 (long arm, q) - 758.330
        17 (long arm, q) - 758.340
        17 (short arm, p) - 758.350
        18 (long arm, q) - 758.340
        18 (short arm, p) - 758.350
        21 (partial or total) - 758.300
        B, NOS - 758.310
        B, NOS - 758.320
        D, NOS (long arm, q) - 758.330
        E (long arm, q) - 758.340
        E (short arm, p) - 758.350
        G, NOS (partial or total) - 758.300
        NOS (unspecified chromosome) - 758.920
        other specified (autosomal) - 758.380
        unspecified (autosomal) - 758.390
        X (partial) - 758.610
Depressions in skull - #754.040
Dermal
        sinus of head - L 744.480
        sinus spine - # 685.100
Dermoid cyst
        epibulbar - L 743.810
        eye - L 743.810
Deviation nasal septum - # 754.020
Dextrocardia
        with complete situs inversus - 759.300
        with situs solitis - 746.800
        without situs inversus - 746.800
Dextroposition
        aorta - 747.260
        heart - see dextrocardia
Diamond-Blackfan syndrome (anemia) - # 284.000
Diaphragm/diaphragmatic
        absent - L 756.600
        elevated - x
        eventration - L 756.620
        hernia
                Bochdalek - L 756.615
                Morgagni - L 756.616
                NOS - L 756.610
                Posterolateral - L 756.615
        other specified anomalies - L 756.680
        paralysis - L 756.680
        unspecified anomalies - L 756.690
Diaphyseal dysplasia, progressive - 756.550
Diastasis recti - x
Diastematomyelia - 742.520
Diastrophic dwarfism - 756.445
Didelphys uterus - 752.200
Diencephalic syndrome - 253.820
DiGeorge syndrome - 279.110
Digestive system, NOS
        absent (complete or partial) - 751.800
        duplication - 751.810
        ectopic - 751.820
        fistula
                with urinary tract - 753.860
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with uterus - 752.320
        obstruction, NOS - 752.900
        other specified anomalies - 751.880
        unspecified anomalies - 751.900
Digit, NOS
        absent - L 755.440
        accessory - see polydactyly
        extra - see polydactyly
        overlapping - L 755.880
Digitalized great toe - L # 755.600
Digitalized thumb - L # 755.500
Dilatation/dilated/dilation - see also large
        aorta - 747.270
       atrium - x
        esophagus - 750.400
        pulmonary artery - 747.330
        pulmonary valve - 746.080
        renal collecting system
                central - L 753.380
                lower - L 753.480
                upper - L 753.480
        renal pelvis - L 753.380
        tricuspid valve - 746.100
        ureter - L 753.220
       vena cava - 747.480
        ventricle (brain) - 742.390
        ventricle (heart) - x
Dimple in chin - x
Disappearing penis syndrome - 752.860
Disease - see syndrome
Dislocatable hip - L 754.310
Dislocation
        elbow - L 754.830
       hip - L 754.300
       knee - L 754.440
        shoulder - x
        tongue - 750.130
Displaced anus - 751.530
Displacement
        cardiac through esophageal hiatus - 750.600
        esophagus - 750.410
        stomach - 750.730
        tongue - 750.130
        uterus - 752.310
Distal arthrogryposis syndrome - L 755.800
Diverticulum
       bladder - 753.820
        esophagus - 750.420
        Meckel's - # 751.010
        stomach - 750.740
        urethral - 753.880
Divisum, pancreas - 751.780
Dolichocephaly - * 754.030
Dorsiflexion of foot - L 754.780
Double - see also duplication
        aortic arch - 747.250
        collecting system (renal) - L 753.410
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inlet left ventricle - 745.300
        inlet right ventricle - 745.300
        kidney (and renal pelvis) - L 753.310
        meatus (urethral, urinary) - 753.840
        ossification center in the manibrium - 756.380
        outlet left ventricle - 745.180
        outlet right ventricle - 745.180
        ureter - L 753.410
        urethra - 753.840
        urethral orifice - 753.840
Double orifice mitral valve - 746.505
Doubling
        cervix - * 752.480
        uterus - 752.200
        vagina - * 752.480
Down syndrome
       facies - 744.910
        karyotype trisomy 21 - 758.000
        karyotype trisomy G, NOS - 758.010
        mosaic - 758.040
        NOS - 758.090
        translocation trisomy (duplication of a 21) - 758.020
        translocation trisomy (duplication of a G, NOS) - 758.030
Downturned mouth - L 744.880
Duane syndrome - # 378.000
Duct
        bile
                agenesis - 751.650
                atresia - 751.650
        hepatic
                agenesis - 751.650
                atresia - 751.650
        omphalomesenteric - 751.000
        vitelline - 751.000
Duodenum
        absent - 751.100
        atresia - 751.100
        stenosis - 751.100
        web - 751.560
Du Pan syndrome - 759.840
Duplex renal collecting system - L 753.410
Duplication - see also double/doubling
        alimentary tract, NOS - 751.810
        chromosome - see also trisomy
                NOS - 758.930
        collecting system (renal) - L 753.410
        digestive system, NOS - 751.810
        esophagus - 750.430
        gallbladder - 751.640
        intestine - 751.500
        nail - L 757.580
        pylorus - 751.500
        renal collecting system - L 753.410
        stomach - 750.750
Dwarf/dwarfism
        Amsterdam - 759.820
        achondroplastic - 756.430
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diastrophic - 756.445
        hypochondrodysplastic - 756.480
        metatrophic - 756.446
        NOS - 756.490
        thanatophoric - 756.447
Dysautonomia, familial - 742.810
Dysgenesis
        adrenal gland - L 759.180
        biliary - 751.670
Dysostosis
        cleidocranial - 755.555
        craniofacial - 756.040
        mandibulofacial - 756.045
        metaphyseal - 756.450
        radioulnar - L 755.536
        spondylocostal - 756.480
Dysmorphic
        aortic valve - 746.480
        mitral valve - 746.505
        pulmonary valve - 746.080
Dysplasia - see also hypoplasia
        aortic valve - 746.480
        bronchopulmonary - x
        caudal - 759.840
        chondroectodermal - 756.520
        cortical (cerebral) - 742.480
        dyssegmental - 756.480
        ears - L * 744.230
        ectodermal
                NOS - 757.340
               other specified - 757.346
               X-linked type - 757.345
        eye - L 743.100
        fronto-nasal - 756.046
        hip
                bilateral - 755.667
               NOS - 755.665
                unilateral - L 755.666
        kidney
                bilateral - 753.000
               NOS - 753.009
               unilateral - L 753.010
        kyphomelic - 756.480
        mitral valve - 746.505
        multiple epiphyseal - 756.570
        nail - L 757.580
        oculoauriculovertebral - 756.060
        pulmonary valve (not hypoplasia) - 746.080
        polystotic fibrous - 756.510
        progessive diaphyseal - 756.550
        pulmonary valve - 746.080
        rib - L 756.340
        Septo-optic - 742.880
        spondyloepiphyseal - 756.460
        spondylometaphyseal - 756.480
        spondylothoracic - 756.480
        Streeter syndrome/dysplasia - # 658.800
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thoracic-pelvic-phalangeal- 756.400
        tricuspid valve - 746.100
Dyssegmental dysplasia - 756.480
Dystrophy/dystrophic
        asphyxiating thoracic - 756.400
        mvotonic - 759.890
        nail - L 757.580
-E-
Eagle-Barrett's syndrome - 756.720
        absent - L 744.010
        absent carilage - L * 744.230
        anomaly NOS - L 744.300
        appendage (not preauricular) - L # 744.120
        asymmetry - x
        bat - L # 744.220
        benign neoplasm - L # 216.200
        cauliflower - L * 744.230
        cleft - L * 744.230
        crease - L 744.280
        decreased cartilage - L * 744.230
        deformity NOS - L 744.300
        dysplastic - L * 744.230
        elfin - L * 744.230
        hypoplastic (not microtia) - L * 744.230
        inner ear anomalies - L 744.030
        large - L 744.200
        lobule (not preauricular) - L # 744.120
        lop - L * 744.230
        low set - L # 744.245
        malformed - L * 744.230
        middle ear anomalies - L 744.020
        misplaced - L 744.240
        other misshapen - L * 744.230
        other specified - L 744.280
        papilloma - L # 744.120
        pit (not preauricular) - L 744.280
        pit (preauricular) - L # 744.210
        pixie-like - L * 744.230
        pointed - L * 744.230
        posteriorly rotated - L # 744.246
        rotated - L # 744.246
        small (not microtia) - L * 744.230
        tag (not preauricular) - L # 744.120
        unspecified anomalies - L 744.300
        unspecified, with hearing impairment - L * 744.090
Ear canal - see auditory canal
Ebstein's anomaly - 746.200
Echogenic kidney - x
Ectodermal dysplasia
        NOS - 757.340
        other specified - 757.346
        X-linked type - 757.345
Ectopia (ectopic) cordia - 746.880
Ectopia vesicae - 753.500
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Ectopic - see also displacement
       adrenal gland - L 759.130
       alimentary tract, NOS - 751.820
       anus - 751.530
       bladder - 753.810
       breast (with accessory nipple) - L 757.620
       digestive system, NOS - 751.820
       heart - 746.880
        kidney - L 753.330
       lung tissues - L 748.600
        nipple
               only - L # 757.650
               with accessory breast - L 757.620
        pancreas - 751.730
        pupil - L 743.440
       spleen - 759.050
       testicle - L 752.530
       ureter - L 753.420
       urethra - 753.850
       urethral orifice - 753.850
Ectrodactyly
       foot - L 755.350
       hand - L 755.250
       NOS - L 755.440
Ectrodactyly-Ectodermal dysplasia-Clefting syndrome - 759.840
Ectropion - L 743.610
Edema
       hereditary, of legs - 757.000
       not of leas - x
Edwards syndrome
       karyotype normal (Edwards phenotype) - 758.295
        karyotype trisomy 18 - 758.200
        karyotype trisomy E, NOS - 758.210
       mosaic - 758.240
       NOS - 758.290
       translocation trisomy 18 (duplication or an 18) - 758.220
       translocation trisomy 18 (duplication or an E, NOS) - 758.230
EEC syndrome - 759.840
Ehlers-Danlos syndrome - 756.850
Eisenmenger's syndrome - 745.410
Elbow
       anomalies - L 755.540
       dislocation - L 754.830
       hyperextension - L 755.540
       webbed - L 755.800
Elevated diaphragm - x
Elfin ear - L * 744.230
Ellis-van Creveld syndrome - 756.525
Elongated - see long
Embryonic remnants (male) cyst - L 752.870
Embryopathia, NEC - 759.910
Emphysema, lobar - L 748.880
Encephalocele
       frontal - 742.085
       frontonasal - 742.085
       occipital - 742.000
       occipitocervical - 742.000
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other specified site - 742.080
        parietal - 742.086
        posterior - 742.000
        sphenoid - 752.080
        unspecified site - 742.090
Encephalocutaneous angiomatosis - 759.610
Encephalopathy, congenital - x
Enchondromatosis - 756.410
Endocardial cushion defect
        NOS - 745.690
        other - 745.680
Endocardial fibroelastosis - 425.300
Endocrine gland
        other specified anomalies - 759.280
        unspecified anomalies - 759.290
Endothelial vessel - L 747.880
Engelmann syndrome - 756.550
Enlarged - see large
Enophalmia - L # 743.800
Enophthalmos - L # 743.800
Enterogenous - 751.500
Entropion - L 743.620
Ependymal cysts - 742.420
Epiblepharon - L * 743.630
Epicanthal folds - L # 743.800
Epidermal nevus syndrome - 757.300
Epidermolysis bullosa - 757.330
Epigastric hernia - 756.795
Epiglottis
        anomalies - 748.300
        hypoplastic - 748.300
Epiloia - 759.500
Epiphyseal dysplasia, multiple - 756.570
Epispadias - 752.610
Epoophoron cyst - L 752.110
Epstein's pearls - x
Epulis - x
Equinovalgus - L 754.680
Equinovarus - L 754.500
Equinus foot - L 754.730
Erb's palsy - L # 767.600
Escobar syndrome - 759.840
Esophagus/esophageal
        atresia
               without tracheoesophageal fistula - 750.300
               with tracheoesophageal fistula - 750.310
        dilatation - 750.400
        displacement - 750.410
        diverticulum - 750.420
        duplication - 750.430
        fistula - 750.480
        giant - 750.400
        other specified anomalies - 750.480
        pouch - 750.420
        short - x
        stenosis - 750.340
        unspecified anomalies - 750.910
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web - 750.350
Esotropia - # 368.000
Ethmocephaly - 759.800
Eustacian tube
        absent - L 744.250
        anomaly - L 744.250
Eustacian valve - x
Eventration of diaphragm - L 756.620
Eversion/everted eyelid - L 743.610
Eversion foot - L 754.680
Exencephaly - 740.020
Exomphalos - 756.700
Exophthalmos - L # 743.800
Exostosis - 756.470
Exotropia - # 378.000
Exstrophy
        bladder - 753.500
        cloaca - 751.550 and 756.790
        lung - L 748.690
External auditory meatal stenosis - L 744.000
External genitalia, absent
        female - * 752.440
        male - 752.880
Extra - see also accessory
        chromosome - see trisomy
        digit - see polydactyly
        finger - see polydactyly
        renal pelvis - L 753.380
        rib
                in cervical region - L # 756.200
               other - L 756.330
        toe - see polydactyly
Extremity - see limb
Extroversion bladder - 753.500
Eye/eyes
        absent - L 743.000
        agenesis - L 743.000
        almond shaped - L # 743.800
        aplasia - L 743.100
        asymmetry - x
        bulging - L # 743.800
        close set - * 756.080
        crossed - # 368.000
        deep set - L # 743.800
        dysplasia - L 743.100
        enlarged - L 743.210
        flat - L # 743.800
        fused
                closed - L * 743.630
                together - 759.800
        Harlequin deformity - L 743.670
        hypoplasia - L 743.100
        mesodermal dysgenesis - L 743.900
        other specified - L # 743.800
        prominent - L # 743.800
        protruding - L # 743.800
        rudimentary - L 743.100
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slant (upward, downward) - L # 743.800
        small - L 743.100
        sunken - L # 743.800
        sun-setting - x
        unspecified - L 743.900
        wide set - 756.085
Eyebrow
        absent - L 744.880
Eyelash
        absent - L * 743.630
        long - L * 743.630
Eyelid
        absent - L * 743.630
        benign neoplasm - L # 216.100
        coloboma - L 743.636
        eversion/everted - L 743.610
        fused - L * 743.630
        other specified - L * 743.630
        weak - L * 743.630
-F-
Face/facial
        absent - L 744.880
        anomaly NOS - 744.910
        asymmetry - 754.000
        asymmetry crying - L 351.000
        benign neoplasm - # 216.300
        broad - 744.910
        cleft - L 744.880
        flat profile - 744.910
        microsomia - L 756.065
        other specified anomalies - L 744.880
        other specified bone anomalies - * 756.080
        palsy - L # 351.000
        skin tag - L # 744.110
        small - 744.910
        teratoma - 238.010
        triangular - 744.910
        unspecified bone anomalies - 756.090
Facies - see also features
        compression - 754.010
        Down syndrome - 744.910
        flat - 744.910
        Potter's - 754.010
Facio-auricular-digital syndrome - 759.800
Facio-auriculo-vertebral syndrome - 756.060
Fallopian tube
        absent - L 752.100
        hypoplastic - L 752.190
        other and unspecified anomalies - L 752.190
Fallot's pentalogy - 745.210
Fallot's tetralogy - 745.200
Familial dysautonomia - 742.810
Fascia
        other specified anomalies - L 756.880
Features
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abnormal - 744.910
        Down syndrome - 744.910
        dysmorphic - 744.910
        Trisomy 21 - 744.910
Female genitalia (external)
        benign neoplasm - # 221.000
        other specified anomalies - * 752.480
        unspecified anomalies - 752.490
Femoral fibular hypoplasia – unusual facies syndrome - 759.840
Femoral hypoplasia - unusual facies syndrome - 759.840
Femur
        absent
                only - L 755.320
                with absent tibia and fibular (total or partial)- L 755.310
                with absent tibia, fibula, and foot - L 755.300
        anteversion - L 755.650
        bowed - L 754.400
        hypoplastic - L 755.650
        other specified anomalies - L 755.650
        short - L 755.650
        torsion - L 755.650
Femur-fibula-ulna syndrome - 759.840
Fenestrated ASD - 745.510
Fetal
        Accutane (Isoretinoin) syndrome - 760.760
        akinesia deformation sequence - 759.840
        alcohol
                effect - 760.720
                syndrome - 760.710
        Dilantin syndrome - 760.750
        hydantoin syndrome - 760.750
FG syndrome - 759.800
Fibroelastosis, endocardial - 425.300
Fibromatosis colli - L 754.100
Fibrosis
        liver - 751.610
        myocardial - 425.300
Fibula
        absent
                only (total or partial) - L 755.366
                with absent femur (total or partial) and tibia (total or partial)- L 755.310
                with absent femur (total or partial), tibia, and foot - L 755.300
                with absent tibia - L 755.320
                with absent tibia and foot - L 755.330
        bowed - L 754.410
        hypoplastic - L * 755.630
        other specified anomalies - L * 755.630
        short - L * 755.630
Fibular hemimelia - L 755.366
Fibular ray defect, NOS - L 755.366
Filum terminale, fat - x
Fimbrial cyst - L 752.120
Finger
        absent
                fifth (with or without fourth) - L 755.270
                first (thumb) - L 755.260
                first (thumb) with absent radius (total or partial) - L 755.260
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NOS - L 755.240
                third (with or without second, fourth) - L 755.250
        acrodactylia - L # 755.500
        anomalies - L # 755.500
        arachnodactyly - L # 755.500
        bifid (thumb) - L 755.010
        brachydactyly - L # 755.500
        camptodactyly - L # 755.500
        clinodactyly - L # 755.500
        club - L 754.840
        cortical (thumb) - x
        digitalized (thumb) - L # 755.500
        flexion deformity - L # 755.500
        fused - L 755.100
        hyperextension - L # 755.500
        hypoplastic
                all other - L 755.585
                thumb (isolated) - L 755.260
        incurving - L # 755.500
        long - L # 755.500
        nubbin - L 755.240
        overlapping - L # 755.500
        rudimentary - L 755.240
        short - L # 755.500
        small, all other - L # 755.500
        small, thumb - L # 755.500
        symbrachydactyly - L # 755.500 and L 755.190-755.199 (depending on the laterality)
        symphalangism - L # 755.500
        syndactyly, unspecified
                bilateral - 755.192
                NOS - 755.193
                unilateral - 755.191
        triphalangeal (thumb) - L # 755.500
        webbed - L 755.110
Fissure
        anal - x
        rectal - * 751.580
        thin palpebral - L 743.635
Fistula
        anal - 751.540
        anourethral - 753.860
        anovaginal - 752.420
        anovesical - 753.860
        branchial cleft - L 744.400
        bronchoesophageal - 750.330
        bronchopulmonary - L 748.350
        digestive tract with uterus - 752.320
        digestive-urinary tract - 753.860
        esophageal - 750.480
        Fourchette - * 752.480
        hepatic artery-portal vein - 747.450
        lip - 750.260
        portal vein-hepatic artery - 747.450
        rectal - 751.540
        rectourethral - 753.860
        rectovaginal - 752.420
        rectovesical - 753.860
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tracheoesophageal
                H type - 750.325
                with esophageal atresia - 750.310
                without esophageal atresia - 750.320
        urethral, NOS - 753.870
        urethrorectal - 753.860
        urinary tract with uterus - 752.320
        uterointestinal - 752.320
        uterovesical - 752.320
        uterus with digestive or urinary tract - 752.320
        vesicovaginal - 752.420
Flat
        eye - L # 743.800
        facial profile - 744.910
        facies - 744.910
        foot - L 754.610
        hand - L 754.880
        head - 754.080
        midface 744.910
        occiput - * 756.080
        side of head - L * 754.050
Flexed wrist - L 755.520
Flexion deformity finger - L # 755.500
Flexion deformity toe - L # 755.600
Fontanelle
        absent - # 754.040
        large - # 754.040
        small - # 754.040
        three - # 754.040
Foot
        abduction - L 754.690
        absent
                only - L 755.340
                with absent femur (total or partial), tibia, and fibula - L 755.300
                with absent lower leg - L 755.330
                with absent tibia and fibula (total or partial) - L 755.330
        adduction - L 754.590
        anomalies - L 755.610
        broad - L 755.610
        claw - L 755.350
        cleft - L 755.350
        clubbed - L 754.730
        deformities, NOS - L 754.735
        dorsiflexion - L 754.780
        ectrodactyly - L 755.350
        equinus - L 754.730
        eversion - L 754.680
        flat - L 754.610
        hyperextended - L 754.780
        hypoplasia - L 755.685
        inversion - L 754.590
        large - L 755.610
        lobster-claw - L 755.350
        long - L 755.610
        oligodactyly - L 755.340
        other specified anomalies - L 754.780
        plantar crease, deep - L 755.610
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plantar furrow - L 755.610
        positional defect, NOS - L 754.780
        rocker-bottom - L # 755.616
        short - L 755.610
        small - L 755.610
        split - L 755.350
        turns
                inward - L 754.590
                outward - L 754.690
                upward - L 754.780
        vertical talus - L # 755.616
Foramina of Magendie and Luschka atresia - * 742.310
Forearm
        absent
                only - L 755.220
                with absent hand - L 755.230
                with absent upper arm - L 755.210
        anomalies - L 755.530
        hemimelia - L 755.230
        short - L 755.530
Forehead
       hirsute - # 744.910
        other anomalies - * 756.080
Forelock, white - # 757.390
Foreskin
        absent - 752.860
        hooded - 752.860
        incomplete - x
        redundant - x
Fossa ovalis atrial septal defect - 745.510
Fourchette fistula - * 752.480
Fragile X syndrome - 758.880
Fragilitas ossium - 756.506
Franceschetti syndrome - 756.045
Frasier syndrome - 759.800
Freeman Sheldon syndrome - 759.800
Frenulum (tongue, lingual)
        anterior - # 750.000
        short - # 750.000
        thick - x
Frenulum (upper lip)
        anomalies - 750.270
        thick - x
Frontal bossing - 754.080
Frontal lobe anomalies - 742.200
Fronto-nasal dysplasia - 756.046
Fryn syndrome - 759.840
Fukuyama congenital muscular dystrophy - 759.890
Funnel chest - 754.810
Fused/fusion
        adrenal glands - L 759.180
        eyes
                closed - L * 743.630
                together - 759.800
        eyelid - L * 743.630
fingers - L 755.100
        kidney - 753.320
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legs - 759.840
        lung lobes - L 748.580
        ossicles (ear)- L 744.020
        penoscrotal - 752.880
        radius and ulna - L 755.536
        rib - L 756.320
        sacroiliac joint - L 755.670
        scrotum - x
        suture
               basilar - 756.030
               coronal - L 756.010
               lambdoidal - L 756.020
               metopic - 756.006
               NOS - 756.000
               other - 756.030
               sagittal - 756.005
        thalami - 742.260
        toes - L 755.120
        ulna and radius - L 755.536
        vertebrae
                cervical - 756.140
               lumbar - 756.160
               NOS - 756.180
               sacral - 756.170
               thoracic - 756.150
        vulva - * 752.440
Galactokinase deficiency - # 271.110
Galactosemia
        classic - # 271.100
        NOS - # 271.190
Gallbladder
        agenesis - 751.630
        duplication - 751.640
        hypoplasia - 751.630
       other anomalies - 751.640
        small - x
Gangliosidosis - # 330.100
Gartner's duct cyst - L 752.110
Gardner syndrome - 759.630
Gastric volvulus - x
Gastroesophageal reflux (GER) - x
Gastroschisis - 756.710
Gaucher disease Type II - 759.870
Genitalia absent (sex unknown) - * 752.790
Genital organs, unspecified anomalies - 752.900
Genu
        recurvatum - L 754.430
       valgum - L 755.645
        varum - L 755.646
Gerbode defect - 745.420
Giant
        esophagus - 750.400
        kidney - L 753.340
Gingiva, cleft - 749.100
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-G-

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Glabella, prominent - # 748.180
Glaucoma - L 743.200
Glioependymal cysts - 742.420
Glossoptosis - 750.130
Glottic web - 748.205
Gluteal cleft, asymmetric - x
Glycogen storage disease - # 271.000
Goiter, congenital - 759.210
Goldenhar syndrome - 756.060
Goltz syndrome - 757.300
Gonadal dysgenesis, pure - 752.720
Gracile rib - L 756.340
Great veins
        other specified anomalies - 747.480
        unspecified anomalies - 747.490
Gum
        cleft - 749.100
        hypertrophy - 750.280
        hypoplastic - x
        other anomalies - 750.280
        prominent gum - 750.280
-H-
Hair
        beaded - 757.410
        Taenzer's - 757.430
       twisted - 757.420
        other specified anomalies - 757.480
        unspecified anomalies - 757.910
        whorl anomalies - #757.390
Hairline
       low anterior - # 744.910
       low NOS - # 744.900
       low posterior - # 744.900
Hairy nevus - *216.920
Hallermann-Streiff syndrome - 756.046
Hallux
        valgus - L 755.605
       varus - L 755.606
Hamartoma
        other specified - 759.680
        unspecified - 759.690
Hammer toe - L # 755.600
Hand
        abnormal position
               with mention of forearm/wrist bone abnormality - L 754.840
               without mention of forearm/wrist bone abnormality- L 755.520
        absent
               only - L 755.240
               with absent forearm - L 755.230
               with absent humerus (total or partial), radius, and ulna - L 755.200
               with absent radius and ulna (total or partial) - L 755.230
        anomalies - L 755.510
       broad - L 755.510
        claw - L 755.250
        cleft - L 755.250
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clenched - L # 755.500
        club - L 754.840
        ectrodactyly - L 755.250
        finger-like (thumb) - L # 755.500
        flat - L 754.880
        hyperflexion - x
        hypoplasia - L 755.585
        large - L 755.510
        lobster-claw - L 755.250
        long - L 755.510
        narrow - x
        oligodactyly - L 755.240
        other specified anomalies - L 754.880
        short - L 755.510
        small - L 755.510
        spade-like - L 754.850
        split - L 755.250
        ulnar deviation - L 755.520
Harelip - see cleft lip
Harlequin deformity of eye - L 743.670
Harlequin fetus - 757.100
Head
        abnormal shape NOS - 754.090
        absent - 740.080
        asymmetric - 754.055
        box shaped - 754.080
        cloverleaf shape - 756.000
        cone shaped - 754.080
        elongated - * 754.030
        enlarged - * 742.400
        flat - 754.080
        flat side of - L * 754.050
        misshapen - 754.090
        small - 742.100
        square - 754.080
        teratoma - 238.010
        tower - 754.080
        triangular shape - 754.070
Heart
        band, anomalous - 746.910
        block - 746.870
        conduction defects - 746.880
        disease
                acyanotic - 746.920
                cyanotic - 746.930
                NOS - 746.990
        displacement through esophageal hiatus - 750.600
        enlarged - * 746.860
        "hole in the heart" - 745.900
        hypoplastic left - 746.700
        hypoplastic NOS - 746.880
        hypoplastic right - 746.882
        large - * 746.860
        murmur - x
        other specified - 746.880
        tumor - 746.880
Heel, prominent - L 755.610
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Hemangioendothelioma liver - L * 228.040
Hemangioma
       intra-abdominal - L * 228.040
       intracranial - * 228.020
       other sites - L * 228.090
       retinal - L * 228.030
       skin and subcutaneous - * 228.010
       unspecified site - * 228.000
       with chondrodysplasia - 756.420
Hemianencephaly - 740.030
Hemiazygos vein anomalies - L 747.650
Hemicephaly - 740.030
Hemidiaphragm - L 756.617
Hemifacial microsomia - L 756.065
Hemihypertrophy - 759.890
Hemimelia, fibular - L 755.366
Hemimelia forearm - L 755.230
Hemimelia tibia - L 755.365
Hemipelvis - L 755.670
Hemivertebra
       cervical - 756.145
       lumbar - 756.165
       NOS - 756.185
       sacral - * 756.170
       thoracic - 756.155
Hemophilia (all types) - # 286.000
Hemorrhage, central nervous system (CNS) - x
Hepatic artery-portal vein fistula - 747.450
Hepatic duct
       agenesis - 751.650
       atresia - 751.650
       other anomalies - 751.670
Hepatic vein
       stenosis - L 747.650
Hepatitis, neonatal
       NOS - # 774.490
       other specified - #774.480
Hepatomegaly - # 751.620
Hepatosplenomegaly - # 751.620 and # 759.020
Hereditary
       edema of legs - 757.000
       trophedema - 757.000
Hermaphroditism, true - 752.700
Hernia
       bladder - 753.820
       diaphragmatic
                Bochdalek - L 756.615
               Morgagni - L 756.616
               NOS - L 756.610
               Posterolateral - L 756.615
        epigastric - 756.795
       hiatal/hiatus - 750.600
       inguinal
               incarcerated - L * 550.100
               with mention of gangrene - L * 550.000
               with obstruction - L * 550.100
               without obstruction without mention of gangrene - L * 550.900
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paraesophageal - 750.600
        umbilical - # 553.100
Herpes simplex, congenital (in utero infection) - # 771.220
Heterotaxy syndrome - * 759.390
Heterotopia pancreas - 751.780
Hiatal/hiatus
        hernia - 750.600
High arched palate - # 750.240
Hip
        abduction - x
        anomalies - L 755.660
        Barlow positive - L 754.310
        benign neoplasm - L # 216.700
        click - x
        crepitus - x
        dislocatable - L 754.310
        dislocation - L 754.300
        dysplasia
               bilateral - 755.667
               NOS - 755.665
               unilateral - L 755.666
        hyperextended - L 755.660
        hypoplasic
               bilateral - 755.667
               NOS - 755.665
               unilateral - L 755.666
        laxity - x
        loose - x
        Ortolani positive - L 754.310
        positive Barlow - L 754.310
        positive Ortolani - L 754.310
        predislocation - L 754.310
        preluxation - L 754.310
        subluxable - L 754.310
        subluxation - L 754.310
        unstable - L 754.310
        webbed - L 755.800
Hirschsprung's disease
        NOS - 751.330
       long-segment - 751.310
       short-segment - 751.320
Hirsutism
       forehead - # 744.910
        other - # 757.450
"Hole in the heart" - 745.900
Holoprosencephaly - 742.260
Holt-Oram syndrome - 759.840
Honeycomb lung - L 748.420
Hooded foreskin - 752.860
Horner syndrome - L 744.880
Horseshoe kidney - 753.320
Humerus
        absent (total or partial)
               only - L 755.220
               with absent radius and ulna - L 755.210
               with absent radius, ulna, and hand - L 755.200
        hypoplastic - L 755.540
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other specified anomalies - L 755.540
        short - L 755.540
Hurler syndrome - 277.510
Hyaline membrane disease - x
Hydatid of Morgagni cyst - L 752.870
Hydranencephaly - 742.320
Hydrocele, congenital - L # 778.600
Hydrocephaly, without spina bifida
        benign external - x
        communicating - 742.380
        ex-vacuo - x
        non-communicating - 742.380
        other - 742.380
        secondary to intraventricular hemorrhage (IVH) or CNS bleed - x
        unspecified, NOS - 742.390
        with spina bifida - see spina bifida
Hydrocolpos - * 752.430
Hydrocytoma - see skin-benign neoplasm
Hydrometrocolpos - * 752.430
Hydromyelia - 742.540
Hydronephrosis - L 753.200
Hydrophthalmos - L 743.200
Hydrops fetalis - # 778.000
Hydrorachis - 742.540
Hydroureter - L 753.220
Hydroureteronephrosis - L 753.200 and L 753.220
Hvmen
        imperforate - * 752.430
        tag - * 752.480
Hyperconvex nail - L 757.580
Hyperextended/hyperextensibility
        arm - L 755.580
        elbow - L 755.540
        finger - L # 755.500
       foot - L 754.780
        hip - L 755.660
        joints - L 755.880
        knee - L 755.640
        leg - L 755.680
        thigh - x
        toe - L # 755.600
Hyperflexion hand - x
Hyperostosis, infantile cortical - 756.530
Hyperpigmentation of skin - # 757.390
Hyperplasia/hyperplastic
        adrenal, congenital
                classical (salt) water - # 255.200
                classical (simple virilizer) - # 255.210
               NOS - # 255.290
               other than 21-OHP deficiency - # 255.240
        kidney - L 753.340
        lung - x
        primary vitreous, persistent - L 743.500
        pulmonary - x
        spleen - # 759.020
Hypertelorism - 756.085
Hypertelorism-hypospadias syndrome - 759.800
```

```
Hypertension, primary pulmonary artery - L * 747.680
Hypertrichosis - # 757.450
Hypertrophic/hypertrophy
        bladder - x
        breast - x
        cardiomyopathy - * 746.860
        clitoris - * 752.450
        gum - 750.280
        kidney - L 753.340
        nail - L 757.510
        pyloric stenosis - 750.510
        thymus - * 759.240
        urethra - x
        ventricle/ventricular (heart) - L * 746.886
        ventricular septum - * 746.860
Hypochondrodysplasia - 756.480
Hypochondrogenesis - 756.480
Hypoglossia - 750.110
Hypoglossia-hypodactylia syndrome - 759.840
Hypoglycemia, idiopathic - # 251.200
Hypognathia - 524.000
Hypomelia
        arm - L 755.585
Hypoparathyroidism, congenital - # 252.100
Hypophosphatasia, congenital - # 275.330
Hypophosphatemic rickets - # 275.330
Hypopigmentation of skin - # 757.390
Hypopituitarism, congenital - #253.280
Hypoplasia/hypoplastic
        adrenal gland - L 759.110
        alae nasae - # 748.180
        aorta - 747.210
        aortic valve - 746.480
        arm - L 755.585
        atrium - 746.887
        bladder - 753.880
        brainstem - 742.280
        breast (with hypoplastic nipple) - L 757.610
        cheek - L 744.880
        colon - 751.520
        ear (not microtia) - L * 744.230
        epiglottis - 748.300
        eye - L 743.100
        fallopian tube - L 752.190
        femur - L 755.650
        fibula - L * 755.630
        finger
               all other - L 755.585
               thumb (isolated) - L 755.260
        foot - L 755.685
        gallbladder - 751.630
        gum - x
        hand - L 755.585
        heart, NOS - 746.880
        hip
                bilateral - 755.667
               NOS - 755.665
```

```
unilateral - L 755.666
humerus - L 755.540
innominate vein - L 747.650
jugular vein - L 747.650
kidney
        bilateral - 753.000
        NOS - 753.009
        unilateral - L 753.010
labia (majora or minora) - * 752.440
larynx - 748.300
left heart syndrome - 746.700
left ventricle - 746.881
leg - L 755.685
lip - # 744.830
lung - L * 748.510
malar - * 756.080
mandible - 524.000
maxillary - * 756.080
mid-facial - * 756.080
mitral valve - 746.505
muscle - L 756.810
nail - L 757.585
nasal bridge - # 748.180
nipple
        only - L * 757.640
        with hypoplastic breast - L 757.610
nose - 748.100
olfactory nerve - 742.270
ovary - L 752.080
pancreas - 751.700
penis - 752.865
pontine - 742.280
pulmonary
        artery - L 747.380
        lung - L * 748.510
        NOS (heart) - 746.995
        valve - 746.000
radius - L 755.530
rib - L 756.340
right heart - 746.882
right ventricle - 746.882
scrotum - L * 752.810
septum pellucidum - 742.210
spleen - 759.010
sternocleidomastoid muscule - L 754.100
supraorbital ridges - * 756.080
testicle - L * 752.810
thalamus - 742.280
thymus - * 759.240
tibia - L * 755.630
toe
        all other - L 755.685
        first - L 755.365
tricuspid valve - 746.100
ulna - L 755.530
umbilical artery - # 747.500
ureter - L 753.210
```

```
ventricle (heart) NOS - 746.883
        vertebrae
                cervical - 756.140
               lumbar - 756.160
               NOS - 756.180
               sacral - 756.170
               thoracic - 756.150
Hypospadias
        with chordee
               balantic - 752.625
                coronal - 752.625
               first degree - 752.625
               glandular - 752.625
               NOS - 752.620
               penile - 752.626
               perineal - 752.627
               scrotal - 752.627
               second degree - 752.626
               subcoronal - 752.625
               third degree - 752.627
        without chordee
               balantic - 752.605
               coronal - 752.605
               first degree - 752.605
               glandular - 752.605
               mild - 752.605
               NOS - 752.600
               penile - 752.606
               perineal - 752.607
               scrotal - 752.607
               second degree - 752.606
               subcoronal - 752.605
               third degree - 752.607
Hypotelorism - * 756.080
Hypothalamus anomalies - 742.220
Hypothyroidism
        congenital - # 243.990
        secondary/tertiary - # 244.800
-l-
Ichthyosiform erythroderma - 757.197
Ichthyosis congenita
        bullous type - 757.115
        other - 757.190
        unspecified - 757.190
        X-linked - 757.196
Ichthyosis vulgaris - 757.195
Icterus - x
lleum
        absent - 751.120
        atresia - 751.120
        stenosis - 751.120
Ilium anomalies - L 755.670
Immotile cilia syndrome - 759.340
Imperforate
        anus
```

```
with fistula - 751.230
                without fistula - 751.240
        hymen - * 752.430
        meatus (urethral, urinary) - 753.630
Incontinentia pigmenti - 757.350
Incurving
        finger - L # 755.500
        toe - L # 755.600
Indeterminate sex NOS - * 752.790
Infantile cortical hyperostosis - 756.530
Infantile myofibromatosis - 759.680
Infantile spasms, congenital - # 345.600
Infantile spinal muscular atrophy - 335.000
Infection, congenital (in utero infection)
        cytomegalovirus (CMV) - # 771.100
        herpes simplex - # 771.220
        human immunodeficiency virus (HIV) - x
        other specified - #771.280
        parvovirus - 771.280
        rubella - 771.000
        syphilis - # 090.000
        TORCH, unspecified - # 771.090
        toxoplasmosis - # 771.200
        varicella - # 052.000
Inferior vermis anomalies - 742.230
Infraorbital crease - L # 743.800
Inguinal hernia
        incarcerated - L * 550.100
        with mention of gangrene - L * 550.000
        with obstruction - L * 550.100
        without obstruction without mention of gangrene - L * 550.900
Iniencephaly
        closed - 740.200
        open - 740.210
        unspecified - 740.290
Innominate artery, aberrant - L 747.640
Innominate vein anomalies - L 747.650
Insufficiency
        aortic valve - * 746.400
        mitral valve - * 746.600
        pulmonary valve - * 746.020
        single atrioventricular valve - 746.900
        tricuspid valve - * 746.105
        truncal valve - 746.900
Integument
        other specified anomalies - 757.800
        unspecified anomalies - 757.990
Intercalary reduction defect
        arm - L 755.210
        leg - L 755.310
        limb, NOS - L 755.410
Interrupted aortic arch - 747.215
Interrupted inferior vena cava - 747.480
Intestine
        adhesion - 751.420
        aganglionosis
                beyond the rectum - 751.310
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involving no more than the anal sphincter and the rectum - 751.320
               total - 751,300
        band - 751.420
        duplication - 751.500
        large
               absent - 751.200
               atresia - 751.200
               malrotation - 751.400
               stenosis - 751.200
        obstruction - x
        other specified anomalies - * 751.580
        small
               absent - 751.190
               absent, with fistula - 751.195
               atresia - 751.190
               atresia, with fistula - 751.195
               malrotation - 751.495
               short - 751.190
               stenosis - 751.190
               stenosis, with fistula - 751.195
       transposition - 751.510
        unspecified anomalies - 751.590
Intracranial cyst - 742.420
Intussusception - x
Inversion
        atrium (heart) - 746.880
       foot - L 754.590
        ventricular - 745.120
Inverted nipples - x
Iris
        absent - L 743.420
        coloboma - L 743.430
       other specified - L 743.440
Ischiopagus conjoined twins - 759.480
Ischium anomalies - L 755.670
Ivemark syndrome - 759.005
-J-
Jackson-Weiss syndrome - 756.046
Jacobsen syndrome - 757.300
Jadassohn-Lewandasky syndrome - 759.890
Jarcho Levin syndrome - 756.480
Jaw
        asymmetry - * 756.080
        size abnormalities - 524.000
        shape abnormalities - * 756.080
Jaw-winking syndrome - 742.800
Jejunal/jejunum
        absent - 751.110
        asymmetric - * 756.080
       atresia - 751.110
        stenosis - 751.110
       web - * 751.580
Jeune syndrome - 756.400
Johansen-Blizzard syndrome - 759.870
Joints, hyperextended - L 755.880
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-K-

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Kabuki syndrome - 759.800
Kalischer's disease - 759.610
Kartagener syndrome (triad) - 759.340
Kast syndrome - 756.420
Kawasaki disease - x
Keratitis-ichthyosis-deafness syndrome - 757.190
Keratoglobus - L 743.220
KID syndrome - 757.190
Kidney - see also renal
        absent
               bilateral - 753.000
               NOS - 753.009
               unilateral - L 753.010
        accessory - L 753.300
        agenesis
               bilateral - 753.000
               NOS - 753.009
               unilateral - L 753.010
        cyst (single) - L 753.100
        cystic dysplasia - L 753.160
        cystic NOS - L 753.180
        double (and pelvis) - L 753.310
        dysplasia
               bilateral - 753.000
               NOS - 753.009
               unilateral - L 753.010
        echogenic - x
        ectopic - L 753.330
        fused - 753.320
        giant - L 753.340
        horseshoe - 753.320
        hyperplastic - L 753.340
        hypertrophy - L 753.340
        hypoplasia
               bilateral - 753.000
               NOS - 753.009
               unilateral - L 753.010
        large - L 753.340
       lobulated - 753.320
        malrotated - L 753.330
        medullary cystic disease
               adult type - 753.150
               juvenile type - 753.140
        medullary sponge kidney - 753.150
        multicystic (dysplasia) - L 753.160
        other specified anomalies - L 753.380
        other specified cystic disease - L 753.180
        pelvic - L 753.330
       polycystic
               adult type - 753.120
               autosomal dominant - 753.120
               autosomal recessive - 753.110
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infantile type - 753.110
               NOS - 753.130
        small
               bilateral - 753.000
               NOS - 753.009
               unilateral - L 753.010
        triple (and pelvis) - L 753.310
        unspecified anomalies - L 753.900
Kinky hair syndrome - 759.870
Klinefelter syndrome
        karyotype 47,XXY - 758.700
        karyotype 48,XXXY - 758.710
        karyotype 48,XXYY - 758.710
        karyotype 49,XXXXY - 758.710
        NOS - 758.790
        other karyotype with additional X chromosomes - 758.710
Klippel-Feil syndrome - 756.110
Klippel-Trenaunay-Weber syndrome - 759.840
Knee
        anomalies - L 755.640
        dislocation - L 754.440
       hyperextended - L 755.640
       laxity - L 754.440
        subluxation - L 754.440
       valgus - L 755.645
        webbed - L 755.640
Koilonychia, congenital - 757.520
Kyphomelic dysplasia - 756.480
Kyphoscoliosis - 756.120
Kyphosis - 756.120
-L-
Labia (minora or majora)
        enlarged - * 752.440
        hypoplastic - * 752.440
        prominent - * 752.440
Lacrimal aparatus/duct
        absent - L 743.640
        cyst - L 743.660
        obstruction - L # 743.650
        other specified - L 743.660
        stenosis - L # 743.650
Ladd's bands - 751.420
Lagophthalmos - x
Lambdoidal suture
        closed - L 756.020
        craniosynostosis - L 756.020
       fused - L 756.020
Lanugo, persistent or excessive - # 757.450
Large - see also dilatation
       adrenal gland - L 759.180
        aorta - 747.270
       atrium - x
       bladder - x
        clitoris - * 752.450
        cornea - L 743.220
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eve - L 743.210
        fontanelle - # 754.040
        foot - L 755.610
        hand - L 755.510
        heart - * 746.860
        kidnev - L 753.340
        labia (minora or majora) - * 752.440
        lips - # 744.820
        liver - # 751.620
        mouth - 744.800
        nail - L 757.510
        penis - 752.880
        pulmonary artery - 747.330
        pulmonary valve - 746.080
        renal pelvis - L 753.380
        septum pellucidum - x
        spleen - # 759.020
        testicle - 752.820
        thymus - * 759.240
        tongue - 750.120
        tricuspid valve - 746.100
        urethra - x
        vena cava - 747.480
        ventricle (brain) - 742.390
        ventricle (heart) - x
        uvula - x
Larsen's syndrome - 755.810
Laryngotracheoesophageal cleft - 748.385
Laryngomalacia - x
Laryngotracheomalacia - x
Larynx/laryngeal
        anomalies of (and supporting cartilage) - 748.300
        cleft - 748.385
        hypoplastic - 748.300
        other specified anomalies - L 748.380
        stenosis
                NOS - 748.300
                subglottic - * 748.310
        stridor - * 748.360
        subglottic stenosis - * 748.310
        unspecified anomalies - 748.390
        web
                glottic - 748.205
                NOS - 748.209
                subglottic - 748.206
Laurence-Moon-Biedl syndrome - 759.820
Laxity
        hip - x
        knee - L 754.440
Left
        atrioventricular valve - see mitral valve
        semilunar valve - see aortic valve
        superior vena cava - 747.410
Left-sided liver - # 751.620
Leg
        absent - L 755.300
        amelia - L 755.300
```

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amputation, NOS - L 755.385
        benign neoplasm - L # 216.700
        bowed lower lea - L 754.410
        bowed, NOS - 754.420
        deformity, NOS - L 754.490
        edema, hereditary - 757.000
        fused - 759.840
        hyperextended - L 755.680
        hypoplasia - L 755.685
        intercalary reduction defect, NOS - L 755.310
        longitudinal reduction defect
                NOS - L 755.360
               postaxial - L 755.366
               preaxial - L 755.365
        lymphedema - 757.000
        other specified anomalies - L 755.680
        other specified reduction defect - L 755.380
        phocomelia, NOS - L 755.310
        positional deformity - L 755.680
        short - L 755.680
        short lower leg - L * 755.630
        single (fused legs, not one absent) - 759.840
        transverse reduction defect, NOS - L 755.385
        unspecified anomalies - L 755.690
        unspecified reduction defect - L 755.390
Lens
        absent - L 743.300
        coloboma - L 743.340
        displaced - L 743.330
        other specified - L 743.380
        spherical - L 743.310
        unspecified - L 743.390
Lenticonus - L 743.380
Leprechaunism - 759.870
Lethal multiple pterygium syndrome - 759.840
Leukoma cornea - L 743.400
Leukonychia, congenital - 757.530
Levocardia
        only - x
        with situs inversus - 759.310
Limb, NOS
        absent - L 755.400
        amelia - L 755.400
        amputation - L 755.420
        intercalary reduction defect - L 755.410
        phocomelia - L 755.410
        upper - see arm
        longitudinal reduction defect
                NOS - L 755.430
               postaxial - L 755.430
               preaxial - L 755.430
        lower - see leg
        other specified anomalies - L 755.880
        other specified reduction defect - 755.480
        transverse reduction defect, NOS - L 755.420
        unspecified anomalies - L 755.900
        unspecified reduction defect - L 755.490
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Limb-body wall complex - 759.840
Lip
        benign neoplasm - # 216.000
        bowed - L 744.880
        cleft
                lateral - 744.800
                with any cleft palate - L 749.200
                        central - 749.220
                        midline - 749.220
                without cleft palate - L 749.100
                        central - 749.120
                        midline - 749.120
        fistula - 750.260
        hpoplastic - # 744.830
        large - # 744.820
        notched - 750.270
        other anomalies - 750.270
        pit - 750.260
        small - # 744.830
        smooth - 750.270
        thin - # 744.830
Lipochondrodystrophy - 277.510
Lipoma
        intra-abdominal organs - L # 214.300
        intrathoracic organs - L # 214.200
        lumbar - # 214.810
        other specified sites - L # 214.800
        paraspinal - # 214.810
        sacral - # 214.810
        skin and cutaneous tissue
                face - # 214.000
                other - # 214.100
        spermatic cord - # 214.400
        unspecified site - # 214.900
Lipomeningocele - see spina bifida
Lipomyelomeningocele - see spina bifida
Lissencephaly - 742.240
Liver
        absent, total or partial - 751.600
        agenesis, total or partial - 751.600
        cyst - 751.610
        cystic disease - 751.610
        fibrocystic disease - 751.610
        enlarged - # 751.620
        fibrosis - 751.610
        hemangioendothelioma liver - L * 228.040
        left-sided - # 751.620
        other anomalies - # 751.620
        transverse - # 751.620
Lobster-claw
        foot - L 755.350
        hand - L 755.250
Lobulated kidney - 753.320
Lobulated spleen - 759.030
Lop ear - L * 744.230
Long
        arm - x
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finger - L # 755.500
        foot - L 755.610
        hand - L 755.510
        head - * 754.030
        neck - # 744.900
        philtrum - 750.270
        skull - * 754.030
        sternum - 756.380
        toe - L # 755.600
Long QT syndrome - 746.880
Longitudinal reduction defect
        arm
                NOS - L 755.265
               postaxial - L 755.270
               preaxial - L 755.260
        leg
               NOS - L 755.360
               postaxial - L 755.366
               preaxial - L 755.365
        limb, NOS
                NOS - L 755.430
               postaxial - L 755.430
                preaxial - L 755.430
Loose hip - x
Lordosis (postural) - 754.210
Loss of chromosomal material - see deletion (chromosome)
Lowe syndrome - 759.870
Lower leg
        absent
               only - L 755.320
               with absent foot - L 755.330
               with absent thigh - L 755.310
        anomalies - L * 755.630
        bowed - L 754.410
        short - L * 755.630
Lower limb - see leg
Low-lying umbilicus - # 759.900
Low set ears - L # 744.245
Lung
        absent - L 748.500
        accessory lobe - L 748.620
        agenesis - L 748.500
        atresia - L 748.500
        bilobar right - 748.625
        cyst
                multiple - L 748.410
               other specified - L 748.480
                single - L 748.400
        cystic adenomatoid malformation - L 748.480
        ectopic tissues - L 748.600
        emphysema, lobar - L 748.880
        exstrophy - L 748.690
        four or more lobes (right) - L 748.620
        fused lobes - L 748.580
        honeycomb - L 748.420
        hyperplasia - x
        hypoplasia - L * 748.510
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incomplete separation of lobes - L 748.580
        lobar emphysema - L 748.880
        lymphangiectasia - L 748.880
        one lobe (left or right) - L 748.580
        other specified anomalies - L 748.690
        other specified dysplasia - L 748.580
        polycystic - L 748.410
        right lung with left lung bronchial pattern - 748.625
        sequestration - L 748.520
        small - L * 748.510
        three lobes (right) - x
        three or more lobes (left) - L 748.620
        two lobes (left) - x
        unspecified anomalies - L 748.690
        unspecified dysplasia - L 748.590
Lutembacher's syndrome - 745.520
Lymphangiectasis of lung - L 748.880
Lymphangioma (any site) - 228.100
Lymphatics, other specified disorders - # 457.800
Lymphedema
        of legs - 757.000
        not of legs - x
-M-
Macrocephaly - * 742.400
Macrocheilia - # 744.820
Macrocolon, not aganglionic - 751.340
Macrocornea - L 743.220
Macrogenitalia (male) - 752.880
Macroglossia - 750.120
Macrognathia - 524.000
Macrostomia - 744.800
Macrotia - L 744.200
Madelung deformity - L * 755.526
Maffucci syndrome - 756.420
Malaligned aorta - 747.260
Malar hypoplasia - * 756.080
Male genitalia (external)
        benign neoplasm - # 222.000
        other specified anomalies - 752.880
Malrotation
        bowel - 751.490
        cecum - 751.400
        colon - 751.400
        kidney - L 753.330
        large bowel - 751.400
        large intestine - 751.400
        midgut - 751.495
        other - 751.490
        small bowel - 751.495
        small intestine alone - 751.495
        unspecified - 751.490
Mandible
        cleft - * 756.080
        hypoplasia - 524.000
Mandibulofacial dysostosis - 756.045
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Manibrium, double ossification center - 756.380
Marble bones - 756.540
Marcus Gunn syndrome - 742.800
Marfan syndrome - 759.860
Maxilla
       hypoplasia - * 756.080
       prominent - * 756.080
Meatus/meatal (external auditory, ear)
       absent - L 744.000
       stenosis - L 744.000
       stricture - L 744.000
Meatus/meatal (urethral, urinary)
       atresia - 753.630
       double - 753.840
       imperforate - 753.630
       obstruction - 753.630
       stenosis - 753.630
Meckel-Gruber syndrome - 759.890
Meckel's diverticulum - # 751.010
Meconium
       peritonitis - # 777.600
       plug syndrome - # 777.100
       stained nails - x
       stained skin - x
Mediastinum cyst - 748.810
Medullary cystic disease kidney
       adult type - 753.150
       juvenile type - 753.140
Medullary sponge kidney - 753.150
Megalencephaly - * 742.400
Megalocolon - 751.340
Megalocornea - L 743.220
Megalogastria - 750.710
Megaloureter - L 753.220
Megameatus - 753.880
Megaurethra - 753.880
Melnick-Fraser syndrome - 759.800
Membranous labyrinth (ear) anomalies - L 744.030
Meningocele - see spina bifida
       cervical - 741.085
       occipital - 742.000
       thoracic - 741.086
       lumbar - 741.087
       sacral - 741.087
Meningomyelocele - see spina bifida
       with unspecified hydrocephalus
               cervical - 741.030
               cervicothoracic - 741.030
               lumbar - 741.050
               lumbosacral - 741.050
               sacral - 741.060
               sacrococcygeal - 741.060
               thoracic - 741.040
               thoracolumbar - 741.040
Menkes syndrome - 759.870
Mermaid syndrome - 759.840
Mesentary anomalies - 751.410
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Mesenteric remnant cyst - L 752.110
Mesocardia - 746.880
Mesodermal dysgenesis eye - L 743.900
Metaphyseal dysostosis - 756.450
Metatarsus
        adductus - L # 754.520
       varus - L # 754.520
Metatrophic dwarfism - 756.446
Metopic suture
        closed - 756.006
        craniosynostosis - 756.006
        fused - 756.006
Microcephalus - 742.100
Microcheilia - # 744.830
Microcolon - 751.520
Microcoria - L 743.440
Microcornea - L 743.410
Microgastria - 750.700
Microgenitalia (male) - 752.880
Microglossia - 750.110
Micrognathia - 524.000
Microgyria - 742.250
Micromelia
       arm - L 755.580
       leg - L 755.680
Micropenis - 752.865
Microphthalmos - L 743.100
Microsomia
        facial - L 756.065
        hemifacial - L 756.065
Microstomia - 744.810
Microtia - L 744.210
        (hypoplastic pinna and absence or stricture of external auditory meatus)
Midface
        flat - 744.910
        hypoplasia - * 756.080
Midgut malrotation - 751.495
Miller-Dieker syndrome - 759.800
Milroy's disease - 757.000
Misshapen
        rib - L 756.310
        skull - 754.090
        speen - 759.030
        sternum - 756.360
Mitral valve
       abnormal - 746.505
        absent - 746.505
        anomaly - 746.505
        atresia - 746.505
        cleft - 746.505
        double orifice - 746.505
        dysmorphic - 746.505
        dysplastic - 746.505
       hypoplasia - 746.505
        insufficiency - * 746.600
        parachute - 746.505
        prolapse - 746.505
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redundant - x
        regurgitation - * 746.600
       small - 746.505
       stenosis - 746.500
       thickened - 746.500
Moebius syndrome - 352.600
Mohr syndrome - 759.800
Mongolian blue spot - x
Mongoloid slant to eyes - L # 743.800
Monilethix - 757.410
Monodactyly
       hand - L 755.250
       foot - L 755.350
Monorchidism - L 752.800
Monosomy G mosaicism - 758.360
Mosaic
        45,X/46,XX (excludes Turner phenotype) - 758.800
       46,XY/47,XXY (excludes Klinefelter phenotype) - 758.820
        49,XXXXY (excludes Klinefelter phenotype) - 758.830
        Down syndrome - 758.040
        Edwards syndrome - 758.240
        Monosomy G - 758.360
        NOS - 758.900
        Patau syndrome - 758.140
        Turner syndrome - 758.610
        XO/XX (excludes Turner phenotype) - 758.810
       XO/XY (excludes Turner phenotype) - 758.800
       XXXXY (excludes Klinefelter phenotype) - 758.830
        XY/XXY (excludes Klinefelter phenotype) - 758.820
       XYY male - 758.840
Mouth
       abnormal shape - L 744.880
       asymmetry - L 744.880
       carp shape - L 744.880
       downturned - L 744.880
       large - 744.800
       lateral cleft - 744.800
       other specified anomalies - 750,280
       small - 744.810
       unspecified anomalies - 750.900
Mucocele - x
Multicystic (dysplasia)
       kidney - L 753.160
       pancreas - 751.780
       renal - L 753.160
Multiple congenital anomalies - 759.700
Multiple epiphyseal dysplasia - 756.570
Multiple pterygium syndrome - 759.840
Muscle
       absent - L 756.810
               pectoralis major - L 756.810
       atrophy, infantile spinal - 335.000
       atrophy (specified muscle) - L 756.880
       hypoplastic - L 756.810
       other specified anomalies - L 756.880
        sternocleidomastoid - see sternocleidomastoid muscule
        unspecified anomalies - 756.900
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Muscle-eye-brain disease - 759.890
Muscular dystrophy, Fukuyama congenital - 759.890
Musculoskeletal system, NOS
        unspecified anomalies - 756.990
Myelocele - see spina bifida
Myelodysplasia - 742.510
Myelomeningocele - see spina bifida
       with unspecified hydrocephalus
               cervical - 741.030
               cervicothoracic - 741.030
               lumbar - 741.050
               lumbosacral - 741.050
               sacral - 741.060
               sacrococcygeal - 741.060
               thoracic - 741.040
               thoracolumbar - 741.040
Myocardial fibrosis - 425.300
Myocardium anomalies - * 746.860
Myofibroma (cardiac) - 425.300
Myofibromatosis, infantile - 759.680
Myopathy, congenital, NOS - L 756.880
Myopia - x
Myotonic dystrophy - 759.890
-N-
Nager syndrome - 756.046
Nail
        absent - L 757.500
       club - L 757.540
       duplication - L 757.580
       dysplastic - L 757.580
       dystrophic - L 757.580
       enlarged - L 757.510
        hyperconvex - L 757.580
       hypertrophic - L 757.510
       hypoplastic - L 757.585
       meconium stained - x
       narrow - L 757.585
       other specified anomalies - L 757.580
       short - x
       small - L 757.585
       unspecified anomalies - 757.920
Nail-patella syndrome - 756.830
Nares
       absent - 748.100
       atresia - L 748.000
       small - # 748.180
Narrow/narrowing
       aorta - 747.210
       biparietal - * 756.080
       bitemporal - * 756.080
       chest - 754.820
       hand - x
       nails - L 757.585
       palate - 750.250
       pulmonary artery - L 747.320
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temporal - * 756.080
       truncal valve - 746.900
Nasal bridge
       broad - # 748.180
       flat - # 748.180
       hypoplasia - # 748.180
       wide - # 748.180
Nasal septum
       absent - # 748.180
       deviation - # 754.020
       perforated - 748.140
Neck
       absent - # 744.900
       anomaly NOS - # 744.900
       benign neoplasm - # 216.400
       broad - # 744.500
       long - # 744.900
       other specified anomalies - L 744.880
        redundant skin folds - # 744.500
       short - # 744.900
       skin folds - # 744.500
       skin tag - L # 744.110
       teratoma - 238.020
       thick - # 744.500
       webbed - # 744.500
       wide - # 744.500
Nephrocalcinosis - x
Nephromegaly - L 753.340
Nephrotic syndrome, congenital - L 753.380
Nervous system
       other specified - 742.880
       unspecified - 742.990
Neu-Laxova syndrome - 759.890
Neurocutaneous melanosis syndrome - 757.300
Neurofibromatosis - 237.700
Neurofibromatosis-Noonan syndrome - 237.700
Nevus - see also skin-benign neoplasm
       blue - see skin-benian neoplasm
       flammeus - # 757.380
       hairy - *216.920
       not elsewhere classified - #757.380
Nipple
       absent
               only - L 757.630
               with absent breast - L 757.600
       accessory
               only - L # 757.650
               with accessory breast - L 757.620
       asymmetric - # 757.680
        ectopic
               only - L # 757.650
               with ectopic breast - L 757.620
        hypoplastic
               only - L * 757.640
               with hypoplastic breast - L 757.610
       inverted - x
       small - L * 757.640
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wide spaced - # 757.680
Noonan syndrome - 759.800
Norrie disease - 759.890
Nose
        absent - 748.100
        accessory - 748.110
        agenesis - 748.100
        asymmetry - # 748.180
        benign neoplasm (external) - # 216.300
        bent - # 754.020
        bifid - 748.120
        broad bridge - # 748.180
        cleft - 748.120
        fissured - 748.120
        flat bridge - # 748.180
        hypoplastic - 748.100
        notched - 748.120
        other specified anomalies - # 748.180
        skin tag - L # 744.110
        small - # 748.180
        tubular - 748.185
        underdevelopment - 748.100
        unspecified anomalies - 748.190
        wide bridge - # 748.180
Nostril
        single - 748.185
        small - # 748.180
Notched lip - 750.270
Nubbin
        finger - L 755.240
       toe - L 755.340
Nuchal folds - # 744.500
Nystagmus - # 379.500
-0-
OAV syndrome - 756.060
Obstruction
        alimentary tract, NOS - 751.900
        biliary - x
        bladder outlet - 753.690
        digestive system, NOS - 751.900
        intestinal - x
        lacrimal - L # 743.650
        meatus (urethral, urinary) - 753.630
        pyloric - 750.580
        ureteropelvic junction - L 753.210
        urethra (anterior) - 753.620
        urinary meatus - 753.630
        ventricular outflow tract (left or right) - 746.880
Obstructive uropathy
       at level of bladder or urethra - 753.690
        unilateral - L 753.290
Occipitocervical encephalocele - 742.000
Occiput
        flat - * 756.080
        prominent - * 756.080
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short - * 756.080
Occult spina bifida - 756.100
Ochoa syndrome - 759.800
Oculoauriculovertebral dysplasia - 756.060
Oculomandibulofacial syndrome - 756.046
Oeis syndrome - 759.890
OFD syndrome, type I - 759.800
Olfactory nerve
       absent - 742.270
       hypoplastic - 742.270
Oligodactyly
       foot - L 755.340
       hand - L 755.240
       NOS - L 755.440
Ollier syndrome - 756.410
Omentum
       adhesion - 751.420
       band - 751.420
Omphalocele - 756.700
Omphalomesenteric duct - 751.000
Ondine's Curse syndrome - x
Onychauxis - 757.515
Opitz G/BBB syndrome - 759.800
Optic disc/nerve
       atrophy - L 743.520
       coloboma - L 743.520
       hypoplastic - L 743.520
       specified anomalies - L 743.520
Oral-facial-digital syndrome, type I - 759.800
Orbit (eye) anomalies - L 743.670
Orofaciodigital syndrome, type II - 759.800
Oro-mandibular-limb hypogenesis syndrome - 759.840
Organ of Corti anomalies - L 744.030
Ortolani positive hip - L 754.310
Ossicles (ear)
       fusion - L 744.020
Osteochondrodysplasia - 756.490
Osteodystrophy
       other specified - 756.580
       unspecified - 756.590
Osteogenesis imperfecta - 756.500
Osteopenia - x
Osteopetrosis - 756.540
Osteopoikilosis - 756.560
Osteoporosis - x
Osteopsathyrosis - 756.505
Ostium primum defect - * 745.600
Ostium secundum defect - 745.510
Oto-palato-digital syndrome - 759.800
Ovary
       absent - L 752.000
       accessory - L 752.020
       agenesis - L 752.000
       cyst
               multiple - L 752.085
               single - L 752.080
       hypoplastic - L 752.080
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other specified anomalies - L 752.080
        streak - L 752.010
        torsion - L 752.080
        unspecified - L 752.090
Overlapping
        digit, NOS - L 755.880
        fingers - L # 755.500
        sutures - x
        toes - L # 755.600
Overriding
        aorta - 747.260
        pulmonary artery - L 747.380
        sutures - x
Ovotestis - 752.700
Oxycephaly - 754.080
-P-
Pachygyria - 742.280
Pachyonychia - 757.516
Palate
        absent
                hard - 749.030
                NOS - 749.090
                soft - 749.070
        anterior - see hard
        cleft
                with cleft lip - see cleft lip with any cleft palate
                without cleft lip
                        hard palate (alone) - L 749.000
                                central - 749.020
                                midline - 749.020
                        NOS (hard/soft not specified) - 749.090
                        soft and hard palate - 749.090
                        soft palate (alone) - L 749.040
                                central - 749.060
                                midline - 749.060
                        submucosal
                                hard - 749.020
                                NOS (hard/soft not specified) - 749.090
                                soft - 749.060
        high arched - # 750.240
        narrow - 750.250
        other anomalies - 750.250
        posterior - see soft
        small - 750.250
Palatoschisis - 749.090
Palmar creases
        abnormal - L # 757.200
        simian - L # 757.200
        transverse - L # 757.200
Palpebral fissures
        narrow - L 743.635
        slanting (up-, down-) - L # 743.800
        small - L 743.635
        thick - x
        thin - L 743.635
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Palsy
        Bell's - L # 351.000
        brachial plexus - L # 767.600
        Erb's - L # 767.600
       facial - L # 351.000
Pancreas
        absent - 751.700
        accessory - 751.710
        agenesis - 751.700
        annular - 751.720
        cyst - 751.740
        divisum - 751.780
        ectopic - 751.730
        heterotopia - 751.780
       hypoplasia - 751.700
       multicystic - 751.780
        other specified anomalies - 751.780
        small - 751.700
        unspecified anomalies - 751.790
Papilloma - see skin-benign neoplasm
Parachute mitral valve - 746.505
Paraesophageal hernia - 750.600
Paralysis
        diaphragm - L 756.680
        vocal cord - x
Parathyroid gland anomalies - 759.230
Parovarian cyst - L 752.120
Partial anomalous pulmonary venous return - 747.430
Partial foramen ovale - * 745.590
Parvovirus infection, congenital - 771.280
Patau syndrome
        karyotype trisomy 13 - 758.100
        karyotype trisomy D, NOS - 758.110
        NOS - 758.190
        mosaic - 758.140
        translocation trisomy 13 - duplication or a 13 - 758.120
        translocation trisomy 13 - duplication or a D, NOS - 758.130
Patella
        absent - L 755.647
        rudimentary - L 755.647
Patent
        ductus arteriosus - * 747.000
        foramen ovale
               NOS - * 745.500
               vs ASD - * 745.590
               vs secundum ASD - * 745.590
        urachus - # 753.700
Pearson syndrome - 759.870
Pectoralis major muscle, absent - L 756.810
Pectus
        carinatum - 754.800
        excavatum - 754.810
        NOS - 754.820
Pelvicaliectasis - L 753.380
Pelvic kidney - L 753.330
Pelviectasis - L 753.380
Pelvis
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anomalies - L 755.670
Pena-Shokier syndrome - L 755.800
Pena-Shokeir II syndrome - 759.840
Penis
       absent - 752.850
       adhesions - 752.860
       aplasia - 752.850
       buried - 752.860
       concealed - 752.860
       disappearing penis syndrome - 752.860
       hypoplastic - 752.865
        large - 752.880
       other anomalies - 752.860
       palmae - 752.860
       small - 752.865
       torsion - 752.860
       webbed - 752.621
Penoscrotal fusion - 752.880
Penoscrotal transposition - 752.880
Penoscrotal web - 752.860
Pentalogy of Cantrell - 759.890
Perforated nasal septum - 748.140
Pericardium anomalies - 746.850
Peripheral arteries, other anomalies - L 747.640
Peripheral pulmonary artery branch stenosis - L * 747.325
Peripheral pulmonary stenosis - L * 747.325
Peripheral vascular system
       other specified anomalies - L * 747.680
       unspecified anomalies - L 747.690
Peripheral veins, other anomalies - L 747.650
Peritoneum
       adhesion - 751.420
       band - 751.420
Peritonitis, meconium - # 777.600
Periventricular cyst - 742.420
Persistent hyperplastic primary vitreous - L 743.500
Persistent omphalomesenteric duct - 751.000
Persistent vitelline duct - 751.000
Pes
       cavus - L 754.700
       planus - L 754.610
       valgus - L 754.615
       varus - L 754.590
Petechiae - x
Peter's anomaly - L 743.440
Peutz-Jegher syndrome - 759.600
Pfeiffer syndrome - 756.057
PHACE syndrome - 759.890
Phalange
       absent (individual)
               finger - L 755.240
               toe - L 755.340
Pharynx/pharyngeal
       other anomalies - L 750.210
       other specified anomalies - 750.280
        pouch - L 750.200
        unspecified anomalies - 750.900
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Phenylketonuria (PKU)
        classic - # 270.100
        hyperphenylalaninemia variant - # 270.110
        NOS - # 270.190
Philtrum
        long - 750.270
        prominent - 750.270
        smooth - 750.270
Phimosis - x
Phlebectasia - L 747.630
Phocomelia
        arm - L 755.210
        leg - L 755.310
        limb, NOS - L 755.410
Pierre-Robin sequence - * 524.080
Pigeon chest - 754.800
Pili torti - 757.420
Pilonidal sinus - # 685.100
Pinna
        absent - L 744.010
        accessory - L # 744.100
        benign neoplasm - L # 216.200
        enlarged - L 744.200
        hypoplastic - L 744.210
        large - L 744.200
Piriform aperature
        atresia - L 748.000
        stenosis - L 748.000
Pit
        auricular - L 744.280
        branchial cleft - L 744.400
        ear - L 744.280
        lip - 750.260
       preauricular - L # 744.410
Pituitary gland anomalies - 759.200
Pixie-like - L * 744.230
Plagiocephaly - L * 754.050
Plantar crease, deep - L 755.610
Plantar furrow - L 755.610
Platycephaly - 754.080
Platyspondyly - * 756.180
Pleura anomaly - L 748.800
Pneumothorax - x
Pointed chin - * 756.080
Pointed ear - L * 744.230
Poland syndrome or anomaly - L 756.800
Polycoria - L 743.440
Polycystic
        kidney
                adult type - 753.120
                autosomal dominant - 753.120
               autosomal recessive - 753.110
               infantile type - 753.110
               NOS - 753.130
        lung - L 748.410
Polydactyly
        digit, NOS - L 755.090
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finger
                Type A - L 755.005
                Type B - L * 755.006
                NOS - L 755.095
                preaxial
                        index finger - L 755.010
                        thumb - L 755.010
                postaxial
                        finger - L 755.005
                        finger vs skin tag - L 755.007
                        NOS - L 755.007
                        skin tag - L * 755.006
        thumb - L 755.010
        toe
                big toe - L 755.030
                NOS - L 755.096
                preaxial - L 755.030
                postaxial - L 755.020
                second toe - L 755.030
Polymicrogyria - 742.250
Polyorchidism - 752.820
Polyotia - L # 744.100
Polyploidy - 758.585
Polysplenia - 759.040
Polystotic fibrous dysplasia - 756.510
Polythelia - L # 757.650
Pontine hypoplasia - 742.280
Porencephalic cyst - * 742.410
Porencephaly - * 742.410
Portal vein
        anomalous termination - 747.440
        hepatic artery fistula - 747.450
Port wine stain - # 757.380
Positional deformity
        arm - L 755.580
        foot - L 754.780
        leg - L 755.680
Posterior encephalocele - 742.000
Posterior fossa cyst - 742.230
Posteriorly rotated ears - L # 744.246
Posterior segment of eye
        specified anomalies - L 743.580
        unspecified anomalies - L 743.590
Posterior urethral obstruction - 753.600
Posterior urethral valves - 753.600
Posterolateral diaphragmatic hernia - L 756.615
Potter's facies - 754.010
Potter's syndrome - 753.000
Potter's sequence - 753.000
Pouch
        esophageal - 750.420
        phayngeal - L 750.200
Prader-Willi syndrome - 759.870
Preauricular
        appendage - L # 744.110
        cyst - L # 744.410
        lobule - L # 744.110
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pit - L # 744.410
        sinus - L # 744.410
        tag - L # 744.110
Predislocation of hip - L 754.310
Preluxation of hip - L 754.310
Premature atrial contractions (PACs) - x
Primary pulmonary artery hypertension - L * 747.680
Primary vitreous, persistent hyperplastic - L 743.500
Proboscis - 748.185
Progressive diaphyseal dysplasia - 756.550
Prolapse
        bladder (mucosa) - 753.830
        mitral valve - 746.505
        tricuspid valve - 746.100
Prominent
        clitoris - * 752.450
        eye - L # 743.800
        glabella - # 748.180
        gum - 750.280
        heel - L 755.610
        labia (minora or majora) - * 752.440
        occiput - * 756.080
        philtrum - 750.270
        prepuce of clitoris - x
        renal pelvis - L 753.380
        tonque - x
        xyphoid process - x
Proptosis - L # 743.800
Prostate
        other anomalies - 752.840
Proteus syndrome - 759.890
Protruding/protuberant
        eye - L # 743.800
        tongue - x
Proximal femoral focal deficiency - L 755.380
Prune belly syndrome - 756.720
Pseudocircumcision - x
Pseudocoarctation of aorta - 747.280
Pseudohermaphroditism
        female - 752.720
        male - 752.710
        NOS - 752.730
Pseudotruncus arteriosus - 747.200
Ptosis - L 743.600
Pterygium colli - # 744.500
Pubis anomalies - L 755.670
Pulmonary/pulmonic
        arteriovenous malformation or aneurysm - L 747.340
        artery
                absent - L 747.300
                absent septum between aorta and - 745.000
                agenesis - L 747.300
                aneurysm - 747.330
                atresia
                        without septal defect - L 747.300
                        with septal defect - L 747.310
                collateral vessel involving (but not involving aorta) - L 747.380
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dilatation - 747.330
                enlarged - 747.330
                hypertension, primary - L * 747.680
                hypoplasia - L 747.380
                narrow - L 747.320
                other specified anomalies - L 747.380
                overriding - L 747.380
                small - L 747.380
                stenosis - L 747.320
                stenosis, branch - L * 747.325
                stenosis, peripheral - L * 747.325
                unspecified anomaly - L 747.390
        hyperplasia - x
        hypoplasia (lung) - L * 748.510
        infundibular stenosis - 746.830
        insufficiency or regurgitation - * 746.020
        NOS (heart)
                atresia - 746.995
                hypoplasia - 746.995
                stenosis - 746.995
        subvalvular stenosis - 746.830
        supravalvular stenosis - L 747.320
        valve
                absent - 746.000
                atresia - 746.000
                bicuspid - 746.080
                dilated - 746.080
                dysmorphic - 746.080
                dysplasia - 746.080
                enlarged - 746.080
                hypoplasia - 746.000
                insufficiency - * 746.020
                other specified anomalies - 746.080
                redundant - 746.080
                regurgitation - * 746.020
                small - 746.000
                stenosis - 746.010
                thickened - 746.080
                unspecified - 746.090
        vein
                atresia - 747.480
                stenosis - 747.480
Punctum lacrimale, absent - L 743.640
Pupil - see also iris
        ectopic - L 743.440
Pyelectasis - L 753.380
Pyelocaliectasis - L 753.380
Pyelon duplex or triplex - L 753.310
Pyloric
        atresia - 751.100
        duplication - 751.500
        obstruction - 750.580
        spasm - # 750.500
        stenosis - 750.510
Pylorospasm - # 750.500
```

-Q-

-R-

```
Rachischisis - see spina bifida
Radial ray defect, NOS - L 755.260
Radioulnar
        dysostosis - L 755.535
        synostosis - L 755.536
Radius/radial
        absent
                only (total or partial) - L 755.260
                with absent humerus (total or partial) and ulna - L 755.210
                with absent humerus (total or partial), ulna, and hand - L 755.200
                with absent thumb - L 755.260
                with absent ulna - L 755.220
                with absent ulna (total or partial) and hand - L 755.230
        deviation of hand/wrist with no mention of radial defect - L 755.520
        deviation of hand/wrist with mention of radial defect - L 754.840
        fused with ulna - L 755.536
        hypoplastic - L 755.530
        other specified anomalies - L 755.530
        short - L 755.530
Ranula - x
Receding chin - 524.000
Rectourethral fistula - 753.860
Rectovaginal fistula - 752.420
Rectovesical - 753.860
Rectum/rectal
        absent
                with fistula - 751.210
                without fistula - 751.220
        atresia
                with fistula - 751.210
                without fistula - 751.220
        fissure - * 751.580
        fistula - 751.540
        short - 751.220
        small - 751.220
        stenosis
                with fistula - 751.210
                without fistula - 751.220
Red cell aplasia - # 284.000
Reduction defect of the brain
        brainstem - 742.280
        other - 742.280
        unspecified - 742.290
Redundant foreskin - x
Redundant
        mitral valve - x
        pulmonary valve - 746.080
        tricuspid valve - 746.100
Reflux
        gastroesophageal (GER) - x
        vesicoureteral - L 753.485
Regurgitation
```

```
aortic valve - * 746.400
        mitral valve - * 746.600
        pulmonary valve - * 746.020
        single atrioventricular valve - 746.900
        tricuspid valve - * 746.105
        truncal valve - 746.900
Rieger's anomaly - L 743.480
Renal - see also kidney
        agenesis, NOS - 753.009
        artery
                absent - L 747.610
                other anomalies - L 747.610
                stenosis - L 747.600
        calculi - L 753.350
        collecting system
                dilation (central) - L 753.380
                dilation (lower) - L 753.480
                dilation (upper) - L 753.480
                duplex - L 753.410
        cyst (single) - L 753.100
        dysplasia, NOS - 753.009
        multicystic (dysplasia) - L 753.160
        pelvis
                dilated - L 753.380
                enlarged - L 753.380
                extra - L 753.380
                other and unspecified obstructive defects - L 753.290
                prominent - L 753.380
Renomegaly - L 753.340
Respiratory system
        anomaly NOS - 748.900
        other specified anomalies - L 748.880
        unspecified anomalies - 748.900
Retina
        aneurysm - L 743.510
        coloboma - L 743.535
        degeneration, peripheral - 362.600
        hemangioma - L * 228.030
        specified anomalies - L 743.510
        unspecified anomalies - L 743.590
Retinitis pigmentosa -362,700
Retractile testicle - x
Retrognathia - 524.000
Reversal
        complete mirror reversal of abdominal organs with normal thoracic organs - 759.330
        complete mirror reversal of all organs - 759.300
        complete mirror reversal of thoracic organs with normal abdominal organs - 759.320
Rhabdomyoma
        heart - * 746.860
        organs other than the heart - 759.680
Rhizomelia
        arm - L 755.540
        extremity NOS - L 755.880
        leg - L 755.650
Rib
        absent - L 756.300
        bifid - L 756.310
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cervical - L # 756.200
        dysplasia - L 756.340
        extra
               in cervical region - L # 756.200
               other - L 756.330
        fused - L 756.320
        gracile - L 756.340
        hypoplastic - L 756.340
        less than 12 - L 756.300
        misshapen - L 756.310
        more than 12 - L 756.330
        other anomalies - L 756.340
        rudimentary - L 756.340
        short - L 756.340
        small - L 756.340
        thin - L 756.340
Rickets, hypophosphatemic - # 275.330
Rieger's anomaly - L 743.480
Rieger syndrome - 759.800
Right
        aortic arch - 747.230
        atrioventricular valve - see tricuspid valve
        lung with left lung bronchial pattern - 748.625
        semilunar valve - see pulmonary valve
        superior vena cava, absent -x
Right sided stomach - 750.730
Riley-Day syndrome - 742.810
Ring
        chromosome
               X - 758.610
        vascular - 747.250
Roberts phocomelia syndrome - 759.840
Robinow syndrome - 759.800
Robin sequence - * 524.080
Rocker-bottom foot - L # 755.616
Roger's disease - 745.400
Rokitansky sequence - 759.890
Rotated ear - L # 744.246
Rubella
        congenital (in utero infection) - 771.000
        syndrome, congenital - 771.00
Rubenstein-Taybi syndrome - 759.840
Rudimentary
        eye - L 743.100
        finger - L 755.240
        patella - L 755.647
        rib - L 756.340
        toe - L 755.340
Russell-Silver syndrome - 759.820
-S-
Sacral/sacrococcygeal/sacrum
        agenesis - 756.170
        anomalies - 756.170
        dimple - # 685.100
        hair tuft - x
```

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hemivertebrae - * 756.170
       mass, NOS - 756.179
       sinus - # 685.100
       teratoma - 238.040
Sacroiliac joint fusion - L 755.670
Saethre-Chotzen syndrome - 756.056
Sagittal suture
       closed - 756.005
       craniosynostosis - 756.005
       fused - 756.005
Salivary glands or ducts, other anomalies - L 750.230
Salmon patches - #757.380
Scalp defects
       aplasia cutis - 757.800
       benign neoplasm - # 216.400
       NOS - 757.800
Scaphocephaly (no mention of craniosynostosis) - * 754.060
Scaphoid abdomen - x
Scapula anomalies - L 755.550
Schinzel-Giedion syndrome - 759.860
Schizencephaly - 742.280
Schwachman Diamond syndrome - 759.870
Scimitar syndrome - L 748.690
Sclera, blue - L * 743.450
Sclerocornea - L 743.410
Scoliosis
       cervical - 754.200
       lumbar - 754.200
       NOS - 754.200
       postural - 754.200
       sacral - 754.200
       thoracic - 754.200
Scrotum
       aplasia - L * 752.810
       bifid - 752.820
       fused - x
       hypoplasia - L * 752.810
       other anomalies - 752.820
       shawl - 752.820
       small - L * 752.810
       underdeveloped/undeveloped - L * 752.810
Seckel syndrome - 759.820
Semilunar valve
       left - see aortic valve
       right - see pulmonary valve
Septal closure - see septal defect
Septal defect (heart)
       aortic - 745.010
       atrial
               NOS - * 745.590
               fenestrated - 745.510
               fossa ovalis - 745.510
               ostium primum - * 745.600
               ostium secundum - 745.510
               other specified - 745.580
               primum - * 745.600
               secundum - 745.510
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vs PFO - * 745.590
        atrioventricular - see atroventricular canal
        auricular - * 745.590
        other - 745.800
        unspecified - 745.900
        ventricular (heart)
               apical - 745.480
               cystalline - 745.480
               malalignment - 745.480
               membranous - 745.480
               mid-muscular - 745.480
               muscular - 745.480
               NOS - 745.490
               other specified - 745.480
               perimembranous - 745.480
                septal - 745.480
               sub-cystalline - 745.480
               type I - 745.480
               type II - 745.480
Septo-optic dysplasia - 742.880
Septum pellucidum
        absent - 742.210
        cavum - x
        enlarged - x
        hypoplasia - 742.210
Sequence - see syndrome
Sequestration lung - L 748.520
Sex chromosome
        additional, NOS - 758.860
               see also trisomy
        mosaic - see mosaic
        other specified anomalies - 758.880
        translocation - see translocation
        trisomy - see trisomy
        unspecified anomalies - 758.890
Shawl scrotum - 752.820
Shield chest - 754.825
Shone's complex - 746.880
Short
        achilles tendon - L 754.720
        arm - L 755.580
        colon - 751.520
        esophagus - x
        extremity NOS - L 755.880
        femur - L 755.650
       fibula - L * 755.630
       finger - L # 755.500
       foot - L 755.610
       forearm - L 755.530
       frenulum (tongue, lingual) - # 750.000
       hand - L 755.510
       humerus - L 755.540
       leg - L 755.680
       lower leg - L * 755.630
        nail - x
        neck - # 744.900
        occiput - * 756.080
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radius - L 755.530
        rectum - 751.220
        rib - L 756.340
        small intestine - 751.190
        sterum - 756.380
        thigh - L 755.650
        tibia - L * 755.630
        toe - L # 755.600
        ulna - L 755.530
        umbilical cord - # 759.900
        ureter - L 753.480
        vagina - 752.410
Shoulder
        anomalies - L 755.550
        benign neoplasm - L # 216.600
        dislocation - x
Sickle cell disease
        SS - # 282.600
        SC - # 282.630
        other - # 282.690
Simian crease - L # 757.200
Single
        atrioventricular valve - 746.900
        atrium - 745.610
        leg (fused, not one absent) - 759.840
        lung cyst
                multiple - L 748.410
                single - L 748.400
        nostril - 748.185
        umbilical artery - # 747.500
        ventricle (heart) - 745.300
Sinus
        branchial cleft - L 744.400
        dermal, of head - L 744.480
        pilonidal - # 685.100
        preauricular - L # 744.410
        sacral - # 685.100
        urachal - # 753.700
Sinus of Valsalva aneurysm - 747.240
Sinus wall (nose) anomalies - 748.130
Sirenomelia - 759.840
Situs ambiguous - * 759.390
Situs inversus
        abdominis - 759.330
        complete, with dextrocardia - 759.300
        thoacis - 759.320
        unspecified - * 759.390
        with levocardia - 759.310
        with sinusitis and bronchitis - 759.340
Sjogren-Larsson syndrome - 757.120
Skeletal dysplasia - 756.490
Skin
        absent - 757.395
        benign neoplasm
                abdominal wall - #216.500
                anus - #216.500
                arm - L # 216.600
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auditory canal, external - L # 216.200
       auricle - L # 216.200
       axillary fold - #216.500
       back - #216.500
       breast - #216.500
       buttock - #216.500
       cheek, external - #216.300
       chest wall - #216.500
        ear - L # 216.200
       eyebrow - #216.300
        eyelid, including canthus - L # 216.100
       face, other and unspecified parts - #216.300
       genital organs
                female - #221.000
                male - # 222.000
        groin - #216.500
       hip - L # 216.700
        leg - L # 216.700
       lip - # 216.000
       neck - # 216.400
       nose, external - #216.300
       other specified sites - L # 216.800
       perianal - # 216.500
       perineum - # 216.500
       pinna - L # 216.200
        scalp - #216.400
       shoulder - L # 216.600
       temple - # 216.300
       trunk - # 216.500
       umbilicus - # 216.500
        unspecified site - # 216.900
cyst - # 757.390
hemangioma - * 228.010
hyperpigmentation - # 757.390
hypopigmentation - #757.390
lipoma
       face - # 214.000
       other - # 214.100
meconium stained - x
other specified anomalies - # 757.390
specified syndromes, not elsewhere classified, involving skin anomalies - 757.300
        cheek - L # 744.110
        ear - L # 744.120
       face - L # 744.110
       finger (postaxial) - L * 755.006
       hymen - * 752.480
       neck - L # 744.110
       nose - L # 744.110
       other - # 757.310
       preauricular - L # 744.110
       unspecified - # 757.310
       vagina - * 752.480
unspecified anomalies - 757.900
asymmetry - 754.055
deformity, NOS - 754.090
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tag

Skull

```
depressions - # 754.040
        elongated - * 754.030
        localized defects - * 756.080
        misshapen - 754.090
        other specified bone anomalies - * 756.080
        other specified deformity (no mention of craniosynostosis) - 754.080
        tower - 754.080
        unspecified bone anomalies - 756.090
Slanting (up-, down-) palpebral fissures - L # 743.800
Small - see also hypoplastic, stenosis
        aorta - 747.210
        aortic valve - 746.300
        auditory canal - L 744.000
        bladder - x
        brainstem - 742.280
        breast - x
        chest - 754.820
        chin - 524.000
        colon - 751.520
        ear (not microtia) - L * 744.230
        face - 744.910
        finger, all other - L # 755.500
        finger, thumb - L # 755.500
        fontanelle - # 754.040
        foot - L 755.610
        gallbladder - x
        hand - L 755.510
        head - 742.100
        kidney
                bilateral - 753.000
                NOS - 753.009
                unilateral - L 753.010
        lips - # 744.830
        iung - L * 748.510
        mitral valve - 746.505
        mouth - 744.810
        nail - L 757.585
        nares - # 748.180
        nipple - L * 757.640
        nose - # 748.180
        nostril - # 748.180
        oral cavity - 744.810
        palate - 750.250
        pancreas - 751.700
        penis - 752.865
        pulmonary artery - x
        pulmonary valve - 746.000
        rectum - 751.220
        rib - L 756.340
        scrotum - L * 752.810
        spleen - 759.010
        stomach - 750.700
        testicle - L * 752.810
        thymus - * 759.240
        trachea - 748.330
        tricuspid valve - 746.100
        umbilical cord - # 759.900
```

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uterus - L 752.380
        uvula - x
        vagina - 752.410
        vena cava (inferior or superior) - 747.400
Smith-Lemli-Opitz syndrome - 759.820
Smith-Magenis syndrome - 759.800
Smooth
        lip - 750.270
        philtrum - 750.270
Sotos syndrome - 759.890
Spade-like hand - L 754.850
Spasms, infantile, congenital - # 345.600
Spermatic cord, torsion - L # 608.200
Sphenoid encephalocele - 752.080
Spherophakia - L 743.310
Sphrintzen syndrome - 759.890
Spina bifida
        aperta - see open lesions
        cystica - see closed lesions
        closed lesions (open vs closed not stated)
                with hydrocephalus
                        other - 741.080
                        with aqueductal stenosis, any site - 741.020
                        with Arnold-Chiari malformation, any site - 741.010
                        with hydrocephalus of late onset, any site - 741.070
                        without Arnold-Chiari malformation or aqueductal stenosis
                                cervical - 741.030
                                cervicothoracic - 741.085
                                lumbar - 741.050
                                lumbosacral - 741.087
                                sacral - 741.060
                                site unknown - 741.090
                                thoracic - 741.040
                                thoracolumbar - 741.086
               without hydrocephalus
                        cervical - 741.910
                        cervicothoracic - 741.980
                        lipomeningocele - 741.985
                        lipomyelomeningocele, any site - 741.985
                        lumbar - 741.930
                        lumbosacral - 741.980
                        sacral - 741.940
                        site unknown - 741.990
                        thoracic - 741.920
                        thoracolumbar - 741.980
        occipital - 742.000
        occulta - 756.100
        open lesions
               with hydrocephalus, any site - 741.000
               without hydrocephalus, any site - 741.900
Spinal cord
        dysplasia - 742.510
        hypoplasia - 742.510
        other specified - 742.580
        tethered - 742.580
        unspecified - 742.910
Spinal dysraphism
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cervical - 756.140
        lumbar - 756.160
        NOS - 756.180
        sacral - 756.170
        thoracic - 756.150
Spinal muscular atrophy, infantile - 335.000
Spine
        unspecified anomalies - 756.190
Spleen
        absent - 759.000
        accessory - 759.040
        cyst - 759.080
        ectopic - 759.050
        enlarged - # 759.020
        hyperplasia - # 759.020
        hypoplasia - 759.010
        lobulated - 759.030
        misshapen - 759.030
        on right in heterotaxy syndrome - 759.050
        other specified anomalies - 759.080
        small - 759.010
        unspecified anomalies - 759.090
Splenomegaly - # 759.020
Split - see also cleft
        hand - L 755.250
        foot - L 755.350
Spondylocostal dysostoses - 756.480
Spondyloepiphyseal dysplasia - 756.460
Spondylolisthesis - 756.130
Spondylometaphyseal dysplasia - 756.480
Spondylothoracic dysplasia - 756.480
Sprengel's deformity - L 755.556
Squamosal craniosynostosis - 756.000
Square cranium - 754.080
Square head - 754.080
Stenosis
        anus
               with fistula - 751.230
                without fistula - 751.240
        aortic - 746.300
        aortic valve - 746.300
        appendix - 751.200
        aqueductal (without spina bifida) - 742.300
        bladder neck - 753.610
               other and unspecified - 753.690
        bronchus - L 748.340
        choanal - L 748.000
        colon - 751.200
        duodenum - 751.100
        esophageal - 750.340
        hepatic vein - L 747.650
        ileum - 751.120
        intestine
               large - 751.200
               small - 751.190
               small, with fistula - 751.195
       jejunum - 751.110
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lacrimal duct - L # 743.650
        larynx (not subglottic) - 748.300
        meatus (urethral, urinary) - 753.630
        meatus (external auditory, ear) - L 744.000
        mitral valve - 746.500
        piriform aperature - L 748.000
        pulmonary
                artery - L 747.320
                artery, branch - L * 747.325
                artery, peripheral - L * 747.325
                infundibular - 746.830
                NOS (heart) - 746.995
                subvalvular - 746.830
                valve - 746.010
                vein - 747.480
        pyloric - 750.510
        rectum
                with fistula - 751.210
                without fistula - 751.220
        renal artery - L 747.600
        subglottic - * 748.310
        subvalvular aortic - 746.300
        subvalvular pulmonary - 746.830
        supra-aortic - 747.220
        supravalvular aortic - 747.220
        supravalvular pulmonary - L 747.320
        trachea - 748.330
        tricuspid valve - 746.100
        truncal valve - 746.900
        ureter - L 753.210
        ureteropelvic junction - L 753.210
        urethral
                anterior - 753.620
                other and unspecified - 753.690
        urinary meatus - 753.630
        vena cava (inferior or superior) - 747.400
Sternocleidomastoid muscule
        absent - L 754.100
        anomalies - L 754.100
        contracture - L 754.100
        hypoplastic - L 754.100
        tumor - L 754.100
Sternum
        absent - 756.350
        bifid - 756.380
        curved - 754.820
        long - 756.380
        misshapen - 756.360
        other anomalies - 756.380
        short - 756.380
        wide - 756.380
Stickler syndrome - 759.860
Stomach
        absent
                with absent GI tract - 750.780
                with rest of GI tract intact - 750.700
        displacement - 750.730
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diverticulum - 750.740
        duplication - 750.750
        other specified anomalies - 750.780
        partial thoracic - 750.600
        right sided - 750.730
        small - 750.700
        transposition - 750.730
        unspecified - 750.920
Stork bite - # 757.380
Strabismus, NOS - # 378.900
Streak ovary - L 752.010
Streeter syndrome/dysplasia - # 658.800
Stricture - see also stenosis
        meatus (external auditory, ear) - L 744.000
        ureter - L 753.210
        urethral - 753.690
Stridor, laryngeal - * 748.360
Sturge-Weber syndrome - 759.610
Subclavian artery, aberrant - L 747.640
Subcoronal hypospadias with chordee - 752.625
Subcoronal hypospadias without chordee - 752.605
Subependymal cyst - 742.420
Subglottic
        stenosis - * 748.310
        web - 748.206
Subluxable hip - L 754.310
Subluxation knee - L 754.440
Subluxation of hip - L 754.310
Sunken eye - L # 743.800
Sun-setting eyes - x
Superior vena cava, right, absent - x
Supernumerary - see accessory, extra
Supraorbital ridges, hypoplastic - * 756.080
Suture
        closed
               basilar - 756.030
               coronal - L 756.010
               lambdoidal - L 756.020
               metopic - 756.006
               NOS - 756.000
               other - 756.030
               sagittal - 756.005
        fused
                basilar - 756.030
               coronal - L 756.010
               lambdoidal - L 756.020
               metopic - 756.006
               NOS - 756.000
               other - 756.030
                sagittal - 756.005
        overlapping - x
        overriding - x
Symblepharon - L * 743.630
Symbrachydactyly fingers - L # 755.500 and L 755.190-755.199 (depending on the laterality)
Symbrachydactyly toes - L # 755.600 and L 755.190-755.199 (depending on the laterality)
Symphalangism finger - L # 755.500
Symphalangism toe - L # 755.600
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Syndactyly (fused vs webbed unspecified)
       fingers
               bilateral - 755.192
               NOS - 755.193
               unilateral - L 755.191
       NOS - L 755.190
       NOS - 755.199
       toes
               bilateral - 755.195
               NOS - 755.196
               unilateral - L 755.194
Syndrome (also anomaly, association, disease, sequence)
       Aarskog syndrome - 759.800
       Acrocallosal syndrome - 759.890
       Adams-Oliver syndrome - 759.840
       Adrenogenital syndrome - # 255.290
       Agnathia formation syndrome - 759.800
       Aicardi syndrome - 759.890
       Alagille syndrome - 759.870
       Albers-Schonberg syndrome - 756.540
       Albright-McCune-Sternberg syndrome - 756.510
       Alport syndrome - 759.870
       Amniotic band syndrome - # 658.800
       Androgen insensitivity syndrome - 257.800
       Angelman syndrome - 759.890
       Antimongolian syndrome - 758.300
       Apert syndrome - 756.055
       Baller-Gerold syndrome - 759.840
       Bart syndrome - 757.330
       Beals syndrome - 759.860
       Beckwith syndrome - 759.870
       Beckwith-Wiedemann syndrome - 759.870
       Beemer Langer syndrome - 759.860
       Blepharophimosis syndrome - 759.800
       Bloom syndrome - 759.890
       BOR syndrome - 759.800
       Bonneville-Ullrich syndrome, NOS - 758.690
       Bourneville's disease - 759.500
       Branchial arch syndrome - 759.800
       Brown syndrome - # 378.000
       Caffev syndrome - 756.530
       Camurati-Engelmann syndrome - 756.550
       Cardio-splenic syndrome - 759.890
       Carpenter syndrome - 759.840
       Cat eye syndrome - 758.580
       Caudal regression syndrome - 759.840
       Cerebro-oculo-facial-skeletal syndrome - 759.890
       CHARGE association - 759.890
       Chediak-Higashi syndrome - 757.300
       Clifford's syndrome - x
       Cockayne syndrome - 759.820
       Coffin-Siris syndrome - 759.800
       COFS syndrome - 759.890
       Congenital contractural arachnodactyly syndrome - 759.860
       Congenital rubella syndrome - 771.000
       Conradi syndrome - 756.575
       Constriction band syndrome - # 658.800
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Cornelia de Lange syndrome - 759.820 Costello syndrome - 759.800 Cri du chat syndrome - 758.310 Crouzon's disease - 756.040 Diamond-Blackfan syndrome (anemia) - # 284.000 Diencephalic syndrome - 253.820 DiGeorge syndrome - 279.110 disappearing penis syndrome - 752.860 distal arthrogryposis syndrome - L 755.800 Down syndrome karyotype trisomy 21 - 758.000 karyotype trisomy G, NOS - 758.010 mosaic - 758.040 NOS - 758.090 translocation trisomy (duplication of a 21) - 758.020 translocation trisomy (duplication of a G, NOS) - 758.030 Duane syndrome - # 378.000 Du Pan syndrome - 759.840 Eagle-Barrett's syndrome - 756.720 Ebstein's anomaly - 746.200 Ectrodactyly-Ectodermal dysplasia-Clefting syndrome - 759.840 Edwards syndrome karyotype normal (Edwards phenotype) - 758.295 karyotype trisomy 18 - 758.200 karyotype trisomy E, NOS - 758.210 mosaic - 758.240 NOS - 758,290 translocation trisomy 18 (duplication or an 18) - 758.220 translocation trisomy 18 (duplication or an E, NOS - 758.230 EEC syndrome - 759.840 Ehlers-Danlos syndrome - 756.850 Eisenmenger's syndrome - 745.410 Ellis-van Creveld syndrome - 756.525 Engelmann syndrome - 756.550 Escobar syndrome - 759.840 epidermal nevus syndrome - 757.300 Facio-auricular-digital syndrome - 759.800 Facio-auriculo-vertebral syndrome - 756.060 Femoral fibular hypoplasia - unusual facies syndrome - 759.840 Femoral hypoplasia – unusual facies syndrome - 759.840 Femur-fibula-ulna syndrome - 759.840 Fetal Accutane (Isoretinoin) syndrome - 760.760 Fetal alcohol syndrome - 760.710 Fetal Dilantin syndrome - 760.750 Fetal hydantoin syndrome - 760.750 FG syndrome - 759.800 fragile X syndrome - 758.880 Franceschetti syndrome - 756.045 Frasier syndrome - 759.800 Freeman Sheldon syndrome - 759.800 Fryn syndrome - 759.840 Gardner syndrome - 759.630 Gaucher disease Type II - 759.870 Gerbode syndrome - 745.420 Goldenhar syndrome - 756.060

Hallermann-Streiff syndrome - 756.046

Goltz syndrome - 757.300

Heterotaxy syndrome - * 759.390

Holt-Oram syndrome - 759.840

Horner syndrome - L 744.880

Hurler syndrome - 277.510

Hypertelorism-hypospadias syndrome - 759.800

Hypoglossia-hypodactylia syndrome - 759.840

hypoplastic left heart syndrome - 746.700

immotile cilia syndrome - 759.340

Ivemark syndrome - 759.005

Jackson-Weiss syndrome - 756.046

Jacobsen syndrome - 757.300

Jadassohn-Lewandasky syndrome - 759.890

Jarcho Levin syndrome - 756.480

Jaw-winking syndrome - 742.800

Jeune syndrome - 756.400

Johansen-Blizzard syndrome - 759.870

Kabuki syndrome - 759.800

Kalischer's disease - 759.610

Kartagener (triad) syndrome - 759.340

Kast syndrome - 756.420

Kawasaki disease - x

Keratitis-ichthyosis-deafness syndrome - 757.190

KID syndrome - 757.190

kinky hair syndrome - 759.870

Klinefelter syndrome

karvotype 47,XXY - 758.700

karyotype 48,XXXY - 758.710

karyotype 48,XXYY - 758.710

karvotype 49,XXXXY - 758.710

NOS - 758,790

other karyotype with additional X chromosomes - 758.710

Klippel-Feil syndrome - 756.110

Klippel-Trenaunay-Weber syndrome - 759.840

Larsen's syndrome - 755.810

Laurence-Moon-Biedl syndrome - 759.820

Lethal multiple pterygium syndrome - 759.840

Limb-body wall complex - 759.840

Long QT syndrome - 746.880

Lowe syndrome - 759.870

Lutemabcher's syndrome - 745.520

Maffucci syndrome - 756.420

Marcus Gunn syndrome - 742.800

Marfan syndrome - 759.860

Meckel-Gruber syndrome - 759.890

Meconium plug syndrome - # 777.100

Melnick-Fraser syndrome - 759.800

Menkes syndrome - 759.870

Mermaid syndrome - 759.840

Miller-Dieker syndrome - 759.800

Milroy's disease - 757.000

Moebius syndrome - 352.600

Mohr syndrome - 759.800

Multiple pterygium syndrome - 759.840

Muscle-eye-brain disease - 759.890

Nager syndrome - 756.046

Nail-patella syndrome - 756.830

Nephrotic syndrome, congenital - L 753.380

Neu-Laxova syndrome - 759.890 Neurocutaneous melanosis syndrome - 757.300 Neurofibromatosis-Noonan syndrome - 237,700 Noonan syndrome - 759.800 Norrie disease - 759.890 OAV syndrome - 756.060 Ochoa syndrome - 759.800 oculoauriculovertebral syndrome - 756.060 oculomandibulofacial syndrome - 756.046 Oeis syndrome - 759.890 OFD syndrome, type I - 759.800 Ollier syndrome - 756.410 Opitz G/BBB syndrome - 759.800 oral-facial-digital syndrome, type I - 759.800 orofaciodigital syndrome, type II - 759.800 Oro-mandibular-limb hypogenesis syndrome - 759.840 other specified syndromes affecting facial appearance - 759.800 associated with short stature - 759.820 involving limbs - 759.840 not elsewhere clasified - 759.890 with metabolic disturbances - 759.870 with other skeletal changes - 759.860 Oto-palato-digital syndrome - 759.800 Patau syndrome karvotype trisomy 13 - 758.100 karyotype trisomy D, NOS - 758.110 mosaic - 758.140 NOS - 758.190 translocation trisomy 13 (duplication or a 13) - 758.120 translocation trisomy 13 (duplication or a D, NOS) - 758.130 Pearson syndrome - 759.870 Pena-Shokier syndrome - L 755.800 Pena-Shokeir II syndrome - 759.840 Peter's anomaly - L 743.440 Peutz-Jegher syndrome - 759.600 Pfeiffer syndrome - 756.057 PHACE syndrome - 759.890 Pierre-Robin sequence - * 524.080 Poland syndrome (anomaly) - L 756.800 Potter's sequence (syndrome) - 753.000 Prader-Willi syndrome - 759.870 Proteus syndrome - 759.890 Prune belly syndrome - 756.720 Rieger's anomaly - L 743.480 Rieger syndrome - 759.800 Riley-Day syndrome - 742.810 Roberts phocomelia syndrome - 759.840 Robinow syndrome - 759.800 Robin sequence - * 524.080 Roger's disease - 745.400 Rokitansky sequence - 759.890 Rubella, congenital syndrome - 771.000 Rubenstein-Taybi syndrome - 759.840 Russell-Silver syndrome - 759.820 Saethre-Chotzen syndrome - 756.056 Schinzel-Giedion syndrome - 759.860

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Schwachman Diamond syndrome - 759.870
       Scimitar syndrome - L 748.690
       Seckel syndrome - 759.820
       Shone's complex - 746.880
       Short rib-polydactyly syndrome - 756.480
       Sjogren-Larsson syndrome - 757.120
       Smith-Lemli-Opitz syndrome - 759.820
       Smith-Magenis syndrome - 759.800
       Sotos syndrome - 759.890
       Sphrintzen syndrome - 759.890
       Stickler syndrome - 759.860
       Streeter syndrome/dysplasia - # 658.800
       Sturge-Weber syndrome - 759.610
       TAR syndrome - 759.840
       Taussig-Bing - 745.100
       Tay-Sachs disease - # 330.100
       Testicular feminization syndrome - 257.800
       Thrombocytopenia-absent radius syndrome - 759.840
       Townes-Brock syndrome - 759.890
       Treacher-Collins syndrome - 756.045
       Turner syndrome
               isochromosome - 758.610
               karyotype 45,X [XO] - 758.600
               mosaic (including XO) - 758.610
               NOS - 758.610
               partial X deletion - 758.610
               ring - 758.610
               variant karyotypes - 758.610
       Uhl's syndrome - 746.882
       VACTERL association - 759.890
       VATER association - 759.890
       Velocardiofacial syndrome (VCFS) - 279.110
       Von Hipple-Lindau syndrome - 759.620
       Von Willebrand syndrome - # 286.400
       Waardenburg syndrome - 759.800
       Walker-Warburg syndrome - 742.880
       Weaver syndrome - 759.890
       Werdnig-Hoffman syndrome - 335.00
       whistling face syndrome - 759.800
       Wiedemann-Beckwith syndrome - 759.870
       Wildervanck syndrome - 756.110
       Williams syndrome - 759.800
       Wilson-Mikity syndrome - x
       Wolff-Hirschorn syndrome - 758.320
       Wolff-Parkinson-White syndrome - 426.705
       Zellweger syndrome - 759.870
Synophrys - L 744.880
Synostosis
       astragaloscaphoid - L 755.620
       cranial - see craniosynostosis
       radioulnar - L 755.536
Svnotia - L 744.240
Syphilis, congenital (in utero infection) - # 090.000
Syringoadenoma - see skin-benign neoplasm
Syringohydromyelia - 742.540
Syringomyelia - 742.540
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Tag - see skin tag
Talipes
       calcaneovalgus - L 754.600
       calcaneovarus - L 754.510
       equinovalgus - L 754.680
       equinovarus - L 754.500
       NOS - L 754.730
Talipomanus - L 754.840
Taenzer's hair - 757.430
Tarsal bones, absent - L 755.340
TAR syndrome - 759.840
Taussig-Bing syndrome - 745.110
Tay-Sachs disease - # 330.100
Teeth, natal - # 520.600
Telecanthus - 756.085
Temporal narrowing - * 756.080
Tendon
       absent - L 756.820
       other specified anomalies - L 756.880
       unspecified anomalies - 756.910
Teratoma
       abdomen - 238.030
       coccygeal - 238.040
       face - 238.010
       head - 238.010
       neck - 238.020
       NOS - 238.000
       other specified - 238.080
       sacral/sacrococcygeal - 238.040
Testicle/testis
       absent - L 752.800
       aplasia - L * 752.810
       appendix - L 752.870
       atrophy - L * 752.810
       ectopic - L 752.530
       hypoplasia - L * 752.810
       in inguinal canal - see undescended
       large - 752.820
       non-palpable - see undescended
       other anomalies - 752.820
       regression - L 752.800
       retractile - x
       small - L * 752.810
       torsion - L # 608.200
       undescended
               bilateral - * 752.514
               NOS - * 752.520
               unilateral - L * 752.500
       vanishing - L 752.800
Testicular feminization syndrome - 257.800
Tethered spinal cord - 742.580
Tetralogy of Fallot
       with ASD - 745.210
       without ASD - 745.200
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Thalami, fused - 742.260
Thalamus, hypoplastic - 742.280
Thanatophoric dwarfism - 756.447
Thick/thickened
        aortic valve - 746.480
        bladder - x
        frenulum - x
        mitral valve - 746.500
        neck - # 744.500
        palpebral fissure - x
        pulmonary valve - 746.080
        tongue - 750.120
        tricuspid valve - 746.100
        urethra - x
        ventricular septum - * 746.860
Thigh
        absent
                with absent lower leg - L 755.310
        hyperextended - x
        short - L 755.650
Thin
        lips - # 744.830
        palpebral fissure - L 743.635
        rib - L 756.340
Thoracic cage
        unspecified anomalies - 756.390
Thoracic-pelvic-phalangeal dysplasia - 756.400
Thorax - see chest
Thrombocytopenia-absent radius syndrome - 759.840
Thumb - see finger
Thymus
        absent - * 759.240
        anomalies - * 759.240
        enlarged - * 759.240
        hypoplastic - * 759.240
        hypertrophy - * 759.240
        small - * 759.240
Thyroglossal cyst - 759.220
Thyroglossal duct anomalies - 759.220
Thyroid gland anomalies - 759.210
Tibia
        absent
                only (total or partial) - L 755.365
                with absent femur (total or partial) and fibula (total or partial)- L 755.310
                with absent femur (total or partial), fibula, and foot - L 755.300
                with absent fibula - L 755.320
                with absent fibula (total or partial) and foot - L 755.330
                with absent first toe (with or without second toe) - L 755.365
        angulation - L * 755.630
        bowed - L 754.410
        hemimelia - L 755.365
        hypoplastic - L * 755.630
        other specified anomalies - L * 755.630
        short - L * 755.630
        torsion - L * 755.630
Tibial ray defect, NOS - L 755.365
Toe
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absent
                fifth (with or without fourth) - L 755.366
                first toe (with or without second toe) - L 755.365
                first toe (with or without second toe) and tibia (total or partial) - L 755.365
                NOS - L 755.340
                third (with or without second, fourth)- L 755.350
                with absent long bone leg - L 755.360
        acrodactylia - L # 755.600
        anomalies - L # 755.600
        arachnodactyly - L # 755.600
        brachydactyly - L # 755.600
        camptodactyly - L # 755.600
        clinodactyly - L # 755.600
        digitalized (great toe) - L # 755.600
        flexion deformity - L # 755.600
        fused - L 755.120
        hammer - L # 755.600
        hyperextension - L # 755.600
        hypoplastic
                all other - L 755.685
                first - L 755.365
        incurving - L # 755.600
        long - L # 755.600
        nubbin - L 755.340
        other specified deformities - L 754.780
        overlapping - L # 755.600
        rudimentary - L 755.340
        short - L # 755.600
        symbrachydactyly - L # 755.600 and L 755.190-755.199 (depending on the laterality)
        symphalangism - L # 755.600
        syndactyly, unspecified
                bilateral - 755.195
                NOS - 755.196
                unilateral - L 755.194
        triphalangeal (geat toe) - L # 755.600
        webbed - L * 755.130
        widely spaced first and second - L # 755.600
Tongue
        absent - 750.100
        cleft - 750.140
        cvst - x
        dislocation - 750.130
        displacement - 750.130
        large - 750.120
        other specified - 750.180
        prominent - x
        protruding - x
        small - 750.110
        thick - 750.120
        tie - # 750.000
        unspecified - 750.190
Tooth, natal - # 520.600
TORCH infection, unspecified - # 771.090
Torsion
        femur - L 755.650
        ovary - L 752.080
        penile - 752.860
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spermatic cord - L # 608.200
       testicle - L # 608.200
       tibia - L * 755.630
Torticollis - L 756.860
Total anomalous pulmonary venous return - 747.420
Tower head - 754.080
Tower skull - 754.080
Townes-Brock syndrome - 759.890
Toxoplasmosis, congenital (in utero infection) - # 771.210
Trabeculated bladder - x
Trachea
       atresia - 748.330
       other anomalies - 748.330
       small - 748.330
       stenosis - 748.330
       unspecified anomalies - 748.390
Tracheomalacia - x
Tracheoesophageal
       fistula
               H type - 750.325
               with esophageal atresia - 750.310
               without esophageal atresia - 750.320
       other anomalies - 750.380
Translocation
       balanced autosomal (in normal individual) - 758.400
       other (autosome) - 758.540
       trisomy 13 - 758.120
       trisomy 18 - 758.220
       trisomy 21 - 758.020
       trisomy D, NOS - 758.130
       trisomy E, NOS - 758.230
       trisomy G, NOS - 758.030
Transposition of
       great arteries
               complete - 745.100
               corrected - 745.120
               incomplete - 745.110
               L- - 745.120
               other - 745.180
               unspecified - 745.190
               with inlet VSD - 745.110
               with muscular VSD - 745.100 and 745.480
               without VSD - 745.100
               with perimembraneous VSD - 745.110
               with VSD - 745.110
       great vessels - see great arteries
       penoscrotal - 752.880
       stomach - 750.730
Transverse liver - # 751.620
Transverse reduction defect, NOS
       arm - L 755.285
       leg - L 755.385
       limb, NOS - L 755.420
Treacher-Collins syndrome - 756.045
Triangular
       face - 744.910
       head shape - 754.070
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Tricuspid valve
        abnormal - 746.100
        aneurysm - 746.100
        atresia - 746.100
        bicuspid - 746.100
        cleft - 746.100
        dilated - 746.100
        dysplastic - 746.100
        enlarged - 746.100
        hypoplasia - 746.100
        incompetence - * 746.105
        insufficiency - * 746.105
        other specified anomalies - 746.100
        prolapse - 746.100
        redundant - 746.100
        regurgitation - * 746.105
        small - 746.100
        stenosis - 746.100
        thickened - 746.100
Trigonocephaly (no mention of craniosynostosis) - 754.070
Trilogy of Fallot - 746.840
Triphalangeal (thumb) - L # 755.500
Triphalangeal (great toe) - L # 755.600
Triploidy - 758.586
Trisomy
        1 - 758.520
        2 - 758.520
       3 - 758.520
       4 - 758.520
        5 - 758.520
       6 - 758.510
       7 - 758.510
        8 - 758.500
        9 - 758.510
        10 - 758.510
        11 - 758.510
        12 - 758.510
        13 - 758.100
        14 - 758.520
        15 - 758.520
        16 - 758.520
        17 - 758.520
        18 - 758.200
        19 - 758.520
        20 - 758.520
        21 - 758.000
        22 - 758.520
        C, NOS - 758.510
        D, NOS - 758.110
        E, NOS - 758.210
        G, NOS - 758.010
        NOS (autosome) - 758.520
        NOS - 758.910
        other total (autosome) - 758.520
        partial (autosome) - 758.530
        XXX female - 758.850
        XYY male - 758.840
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Trophedema, hereditary - 757.000
Truncal valve - 746.900
        insufficiency - 746.900
        narrow - 746.900
        regurgitation - 746.900
        stenosis - 746.900
Truncus arteriosus - 745.000
Tuberous sclerosis - 759.500
Tubular hypoplasia of aorta - 747.210
Tumor
        heart - 746.880
        sternocleidomastoid muscule - L 754.100
Turner syndrome
        isochromosome - 758.610
        karyotype 45,X [XO] - 758.600
        mosaic (including XO) - 758.610
        NOS - 758.610
        partial X deletion - 758.610
        ring - 758.610
        variant karyotypes - 758.610
Turricephaly - 754.080
Twin reversed arterial perfusion (TRAP) sequence - 759.890
Twins
        acardiac - 759.480
        conjoined
                craniopagus (head-joined twins) - 759.410
                dicephalus (two heads) - 759.400
                ischiopagus - 759.480
                other specified - 759.480
                pelvis-joined twins - 759.480
                pygophagus (buttock-joined twins) - 759.440
                thoracopagus (thorax-joined twins) - 759.420
                unspecified - 759.490
                xiphopagus (xiphoid-joined twins) - 759.430
Twisted hair - 757.420
Two vessel umbilical cord - # 747.500
Tympanic membrane anomalies - L 744.020
-U-
Uhl's disease - 746.882
Ulna/ulnar
        absent
                only (total or partial) - L 755.270
                with absent humerus (total or partial) and radius - L 755.210
                with absent humerus (total or partial), radius, and hand - L 755.200
                with absent radius - L 755.220
                with absent radius (total or partial) and hand - L 755.230
        bowed without Madelung deformity - L 755.530
        deviation of hand/wrist with no mention of ulnar defect - L 755.520
        deviation of hand/wrist with mention of ulnar defect - L 754.840
        fused with radius - L 755.536
        hypoplastic - L 755.530
        other specified anomalies - L 755.530
        short - L 755.530
Ulnar ray defect, NOS - L 755.270
Umbilical artery hypoplasia - # 747.500
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Umbilical cord/umbilicus
        anomalies - # 759.900
        atrophy - # 759.900
        benign neoplasm - # 216.500
        Four vessel - L * 747.680
        hernia - # 553.100
        low-lying - # 759.900
        short - # 759.900
        single artery - # 747.500
        small - # 759.900
        two vessels - # 747.500
Underdevelopment
        nose - 748.100
Undescended testicle
        bilateral - * 752.514
        NOS - * 752.520
        unilateral - L * 752.500
Unicornate uterus - L 752.380
Unstable of hip - L 754.310
Upper
        alimentary tract
                other specified anomalies - 750.800
                unspecified anomalies - 750.990
        arm
                absent
                        only - L 755.220
                        with absent forearm - L 755.210
                anomalies - L 755.540
        leg - see also thigh
                anomalies - L 755.650
        limb - see arm
Urachus/urachal
        cyst - 753.710
        other and unspecified anomaly - 753.790
        patent - # 753.700
        remnant - 753.790
        sinus - # 753.700
Ureter
        absent - L 753.400
        accessory - L 753.410
        atresia - L 753.210
        dilated - L 753.220
        double - L 753.410
        ectopic - L 753.420
        hypoplastic - L 753.210
        other and unspecified obstructive defects - L 753.290
        other specified anomalies - L 753.480
        short - L 753.480
        stenosis - L 753.210
        stricture - L 753.210
        unspecified anomalies - L 753.910
Ureterectasis - L 753.220
Ureterocele - L 753.480
Ureteropelvic junction
        obstruction - L 753.210
        stenosis - L 753.210
Ureterovesical junction - see ureteropelvic junction
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Urethra/urethral
        absent - 753.800
        anterior
                atresia - 753.620
                obstruction - 753.620
                stenosis - 753.620
                valve - 753.620
        diverticulum - 753.880
        double - 753.840
        ectopic - 753.850
        enlarged - x
        fistula, NOS - 753.870
        hypertrophy - x
        obstruction (posterior) - 753.600
        orifice
                ectopic - 753.850
        other and unspecified atresia and stenosis - 753.690
        other specified anomalies - 753.880
        stricture - 753.690
        thickened - x
        unspecified anomalies - 753.930
        valves (posterior) - 753.600
Urethrorectal fistula - 753.860
Urinary meatus
        atresia - 753.630
        double - 753.840
        obstruction - 753.630
        stenosis - 753.630
Urinary system/tract
        fistula with digestive system - 753.860
        unspecified anomalies - 753.990
Urogenital sinus malformation - 753.880
Uropathy, obstructive
        at level of bladder or urethra - 753.690
        unilateral - L 753.290
Urticaria pigmentosa - 757.320
Uterointestinal fistula - 752.320
Uterovesical fistula - 752.320
Uterus
        absent - 752.300
        agenesis - 752.300
        bicornate - L 752.380
        didelphys - 752.200
        displaced - 752.310
        doubling - 752.200
        fistula (with digestive or urinary tract) - 752.320
        other anomalies - L 752.380
        septate - L 752.380
        small - L 752.380
        unicornate - L 752.380
        unspecified anomalies - 752.390
Uvula
        absent - 749.080
        bifid - 749.080
        cleft - 749.080
        enlarged - x
        small - x
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VACTERL association - 759.890
Vagina
        absent (complete or partial) - 752.410
        agenesis (complete or partial) - 752.410
        atresia (complete or partial) - 752.410
        cyst
                embryonal - # 752.460
                other - 752.470
        doubling - * 752.480
        other specified anomalies - * 752.480
        short - 752.410
        small - 752.410
        tag - * 752.480
        unspecified anomalies - 752.490
Vaginocele - * 752.480
Valga/valgum/valgus
        coxa - L 755.660
        cubitus - L 755.540
        genu - L 755.645
        hallux - L 755.605
        knee - L 755.645
        other specified deformities of foot - L 754.680
        pes - L 754.615
        unspecified deformities of foot - L 754.690
Valve
        aortic
                absent - 746.480
                atresia - 746.480
                bicuspid - * 746.400
                dysmorphic - 746.480
                dysplastic - 746.480
                hypoplastic - 746.480
                incompetence - * 746.400
                insufficiency - * 746.400
                other specified - 746.480
                quadricuspid - 746.480
                regurgitation - * 746.400
                small - 746.300
                stenosis - 746.300
                thickened - 746.480
                unspecified - 746.490
        mitral
                absent - 746.505
                anomaly - 746.505
                atresia - 746.505
                cleft - 746.505
                dysmorphic - 746.505
                dysplastic - 746.505
                hypoplasia - 746.505
                insufficiency - * 746.600
                parachute - 746.505
                prolapse - 746.505
                redundant - x
                regurgitation - * 746.600
```

```
small - 746.505
                stenosis - 746.500
                thickened - 746.500
        pulmonary
                absent - 746.000
                atresia - 746.000
                bicuspid - 746.080
                dilated - 746.080
                dysmorphic - 746.080
                dysplasia - 746.080
                enlarged - 746.080
                hypoplasia - 746.000
                insufficiency - * 746.020
                other specified anomalies - 746.080
                redundant - 746.080
                regurgitation - * 746.020
                small - 746.000
                stenosis - 746.010
                thickened - 746.080
                unspecified - 746.090
        tricuspid
                abnormal - 746.100
                aneurysm - 746.100
                atresia - 746.100
                bicuspid - 746.100
                cleft - 746.100
                dilated - 746.100
                dysplastic - 746.100
                enlarged - 746.100
                hypoplasia - 746.100
                incompetence - * 746.105 insufficiency - * 746.105
                other specified anomalies - 746.100
                prolapse - 746.100
                redundant - 746.100
                regurgitation - * 746.105
                small - 746.100
                stenosis - 746.100
                thickened - 746.100
                unspecified anomalies - 746.900
Vanishing testicle - L 752.800
Varix- L 747.630
Vara/Varum/varus
        complex deformities - L 754.530
        coxa - L 755.660
        genu - L 755.646
        hallux - L 755.606
        metatarsus - L # 754.520
        unspecified (of feet) - L 754.590
Varicella, congenital (in utero infections) - # 052.000
Vascular ring - 747.250
Vas deferens
        atresia - L 752.830
        other anomalies - 752.840
VATER association - 759.890
Vein of Galen anomalies - L 747.810
Velocardiofacial syndrome (VCFS) - 279.110
```

```
Vena cava
        absent (except left superior) - 747.480
        bilateral inferior - 747.480
        bilateral superior - 747.410
        dilated - 747.480
        enlarged - 747.480
        interrupted inferior - 747.480
        left superior - 747.410
        small (inferior or superior) - 747.400
        stenosis (inferior or superior) - 747.400
Ventri in version - 745.120
Ventricle/ventricular (brain)
        cyst - * 742.485
        dilatation - 742.390
        enlarged - 742.390
Ventricle/ventricular (heart)
        common - 745.300
        dilatation - x
        double inlet left - 745.300
        double inlet right - 745.300
        double outlet left - 745.180
        double outlet right - 745.180
        enlarged - x
        hypertrophy - L * 746.886
        hypoplastic left - 746.881
        hypoplastic NOS - 746.883
        hypoplastic right - 746.882
        inversion - 745.120
outflow tract obstruction (left or right) - 746.880
        septal defect
                apical - 745.480
                cystalline - 745.480
                hypertrophy - * 746.860
                malalignment - 745.480
                membranous - 745.480
                mid-muscular - 845.480
                muscular - 745.480
                NOS - 745.490
                other specified - 745.480
                perimembranous - 745.480
                septal - 745.480
                sub-cystalline - 745.480
                thickened - * 746.860
                type I - 745.480
                type II - 745.480
        single - 745.300
Ventriculomegaly - 742.390
Vermian atrophy - 742.230
Vermis (inferior) anomalies - 742.230
Vertebra
        cervical
                agenesis - 756.146
                anomalies - 756.140
                bifid - 756.140
                butterfly - 756.140
                cleft - 756.140
                fused - 756.140
```

```
hypoplastic - 756.140
                segmentation anomalies - 756.140
        lumbar
                agenesis - 756.166
                anomalies - 756.160
                bifid - 756.160
                butterfly - 756.160
                cleft - 756.160
                fused - 756.160
                hypoplastic - 756.160
                segmentation anomalies - 756.160
        NOS
                bifid - 756.180
                butterfly - 756.180
                cleft - 756.180
                fused - 756.180
                hypoplastic - 756.180
                other specified anomalies - 756.180
                segmentation anomalies - 756.180
                unspecified anomalies - 756.190
        sacral/sacrum
                agenesis - 756.170
                anomalies - 756.170
                bifid - 756.170
                butterfly - 756.170
                cleft - 756.170
                fused - 756.170
                hypoplastic - 756.170
                segmentation anomalies - 756.170
        thoracic
                agenesis - 756.156
                anomalies - 756.150
                bifid - 756.150
                butterfly - 756.150
                cleft - 756.150
                fused - 756.150
                hypoplastic - 756.150
                segmentation anomalies - 756.150
Vertical talus foot - L # 755.616
Vesicoureteral reflux - L 753.485
Vesicovaginal fistula - 752.420
Vesiculobullous dermatosis - x
Vitelline duct - 751.000
Vitreous humor anomalies - L 743.500
Vocal cord paralysis - x
Volvulus
        gastric - x
        intestinal - x
Von Hipple-Lindau syndrome - 759.620
Von Willebrand disease - # 286.400
Vulva
        absent - * 752.440
        cyst - 752.470
        fused - * 752.440
        other anomaly - * 752.440
```

```
Waardenburg syndrome - 759.800
Walker-Warburg syndrome - 742.880
Weaver syndrome - 759.890
Web/webbed
       duodenal - 751.560
       elbow - L 755.800
       esophageal - 750.350
       fingers - L 755.110
       hip - L 755.800
       jejunal - * 751.580
       knee - L 755.640
       larynx
               glottic - 748.205
               NOS - 748.209
               subglottic - 748.206
       neck - # 744.500
       penis - 752.621
       penoscrotal - 752.860
       toes - L * 755.130
Werdnig-Hoffman disease - 335.000
Werner mesomelic dysplasia - 756.480
Wharton duct cyst - x
Whistling face syndrome - 759.800
White forelock - # 757.390
Widely spaced first and second toes - L # 755.600
Widely spaced nipples - # 757.680
Wide neck - # 744.500
Wide set eyes - 756.085
Wide sternum - 756.380
Wiedemann-Beckwith syndrome - 759.870
Wildervanck syndrome - 756.110
Williams syndrome - 759.800
Wilson-Mikity syndrome - x
Wolff-Hirschorn syndrome - 758.320
Wolff-Parkinson-White syndrome - 426.705
Wolffian duct cyst - L 752.870
Wrist
       anomalies - L 755.520
       flexed - L 755.520
       ulnar deviation - L 755.520
-X-
Xeroderma pigmentosum - 757.360
XK aprosencephaly - 759.800
Xyphoid process
       bifid - 756.380
       prominent - x
-Y-
-Z-
Zellweger syndrome - 759.870
```

-KARYOTYPES-

```
45.X - 758.600
45.X.inv(9) - 758.600
45,X/46,X+mar - 758.610
45,X/46,X,9(X)(q10) - 758.610
45.X/46.X.r - 758.610
45,X/46,X,r(X)(p22.3;q24) - 758.610
45,X/46,XX (with Turner syndrome phenotype) - 758.610
45,X/46,XX (without Turner syndrome phenotype) - 758.810
45,X/46,XX/46,X,+15/47,XX+15 - 758.520
45,X/46,XX/46,X,+15/47,XX+15 - 758.880
45,X/46,XY - 758.800
45,X/46X,r(X) - 758.610
45,X/47,XXX - 758.610
45,XX,der(14;21)(q10;q10)/46,XX,der(14;21)(q10;q10),+21 - 758.040
45,XX,der(14;21)(q10;q10)/46,XX,der(14;21)(q10;q10),+21 - 758.020
45,XX,der(5)(5;15)(p15.3;q13),-15.15Hder(5)(PML+D15S10-,141-,D5S23+74+) - 758.540
45,XX,der(7)+(7;21)(q35;q10)-21 - 758.540
45,XX,der,(16;22)(p13.3;q11,2),-22 - 758.380
45,XY,?dic(7;20)(p22;?p13)/46,XY,?dic(7;20)(p22;?p13),+mar - 758.540
46 XY,+21,der(21;21)(q10;q10) - 758.020
46,del(13q) - 758.330
46,X,del(X)(p22.2) - 758.610
46,X,i(Xq) - 758.610
46,X,inv(Y)+mar - 758.580
46,XX,+21,der(21;21) - 758.020
46,XX,+21,der(21;21)(q10;q10) - 758.020
46,XX,-14,+t(13;14)/45XX,-14,+t(13;14) - 758.120
46,XX,-18,+der(18)+(18;?)(q12.3;?),var(14) - 758.530
46,XX,-18,+der(18)+(18;?)(q12.3;?),var(14) - 758.340
46,XX,-20,+mar - 758.580
46,XX,-21,+t(21q;21q) - 758.020
46,XX,?del(15)(q11.2)/47,XX+mar/46,XX - 758.380
46,XX,?del(15)(q11.2)/47,XX+mar/46,XX - 758.580
46,XX,9,qh+ - 758.580
46,XX,add(4)(p16) - 758.530
46,XX,add(4)(p16) - 758.530
46,XX,add(6)(p15.1) - 758.530
46,XX,add(8)(p23) - 758.530
46,XX,del(1)(p22) - 758.380
46,XX,del(1)(p36.3) - 758.380
46,XX,del(1)(p36.3),inv(9)(p11;q12)ish del(1)(P36.3)(P58-,D1Z2) - 758.380
46,XX,del(11)(q23) - 758.380
46,XX,del(14;21)(q10;q10)mat,+21 - 758.020
46,XX,del(15)(q11.2;q13) - 758.380
46,XX,del(17)(p11.2;p13) - 758.380
46,XX,del(18)(p11.2) - 758.350
46,XX,del(18)(q?21.1) - 758.340
46,XX,del(22)(q11.2;q11.2) - 758.380
46,XX,del(22)(q11.2;q11.2)(D22575-) - 758.380
46,XX,del(22)(q11.2;q11.2)(D22S75) - 758.380
46,XX,del(3)(g23;g25 or g25;g26.2)DISH del(3)(WCP3+) - 758.380
46,XX,del(4)(q32.1) - 758.380
46,XX,del(5)(p14) - 758.310
46,XX,del(9)(p22) - 758.380
46,XX,del(9)(p22-pter) - 758.380
46,XX,der(?18)t(13;18)(?q11;?p11.1).ISH 46,XX,der(18)t(13;18)(q12;p11.2)(D18Z1+) - 758.120
```

```
46,XX,der(13)+(2;13)(q37.1;q32.2) - 758.540
46,XX,der(13)t(13;?)(q;?) - 758.330
46,XX,der(13)t(13;?)(q;?) - 758.530
46,XX,der(14)+(14;17)(p12;p11.2)pat.ISH der(14)+(14;17)(p12;p11.2)(D17S29-)pat - 758.380
46,XX,der(14)+(14;17)(p12;p11.2)pat.ISH der(14)+(14;17)(p12;p11.2)(D17S29-)pat - 758.530
46.XX.der(14:21)(a10:a10).+21 - 758.020
46,XX,der(15)t(15.15)(p13;q26.1) - 758.580
46,XX,der(21)+(5:21) - 758.530
46,XX,der(21)+(5:21) - 758.540
46,XX,der(21:21)(q10;q10),+21 - 758.020
46,XX,der(5)+(5;10)(p15.1;p11.21).ISH der(5)+(5;10)(p15.1;p11.21)WCP 10+,D5S23-) - 758.540
46,XX,der(7) - 758.580
46,XX,der(8p) - 758.580
46,XX,dup(4)(q28;q33) - 758.530
46,XX,dup(5)(q11.2;q12) - 758.530
46,XX,inv(2) - 758.580
46,XX,inv(3)(?p13;?q21) - 758.580
46,XX,inv(6)(p21.3;q15)pat - 758.580
46,XX,inv(9)(p11;q12),r(13)(p11.2;q22)/45,XX,inv(9)(p11;q12),-13 - 758.380
46,XX,inv(9)(p12;q13) - 758.580
46,XX,inv(9),(p11;q13) - 758.580
46,XX,inv(9),(p12;q13) - 758.580
46,XX,ish del(15)(q11.2;q11.2)(SNRPN-) - 758.380
46,XX male - 758.880
46,XX,r(22)(p11.2;q13.3) - 758.580
46,XX,r(8) - 758.580
46,XX,rob(21q;21q) - 758.020
46,XX,t(11;12)(q22.1;q23) - 758.400
46,XX,t(14;21)(q10;q10),+21 - 758.020
46,XX,t(14q;21q) - 758.020
46,XX,t(21;21)(q10;q10),+21 - 758.020
46,XX,t(21q;21q) - 758.020
46,XX,t(6;7)(p22.2;15.3) - 758.540
46,XX,t(9:13)(q22;q14)pat - 758.400
46,XX.ISH del(22)(q11.2;q11.2)(D22S75-) - 758.380
46,XX.ISH del(22)(q11.2;q11.2)(F5-)/46,X,fra(X)(q27.3).ISH del(22)(q11.2;q11.2)(F5-_ - 758.880
46,XX.ISH del(22)(q11.2;q11.2)(F5-)/46,X,fra(X)(q27.3).ISH del(22)(q11.2;q11.2)(F5-_ - 758.380
46,XX.ISH del(22q11.2;q11.2)(D22575-) - 758.380
46,XX/45,X - 758.610
46,XX/46,XX,fra(X)(q28) - 758.880
46,XX/47,XX,+13 - 758.100
46,XX/47,XX,+21 - 758.040
46,XXI(18)(q10) - 758.220
46,XY,+13,der(13;13)(q10;q10) - 758.120
46,XY,+13,der(13;14)(q10;q10) - 758.120
46,XY,+21,der(21;21)(q10;q10) - 758.020
46,XY,+21,der(21;21)(q10;q10)de novo - 758.020
46,XY,-10,der(10)t(3;10)(p25;q26)mat - 758.400
46,XY,-14,+der(14)rob(13q;14q) - 758.120
46,XY,-14,+t(13:14)(p11;q11) - 758.120
46,XY,-14,+t(14q;21q) - 758.020
46,XY,-21,+der(21) - 758.000
46,XY,-21,+t(21q;21q) - 758.020
46,XY,?del(7)(q36) - 758.380
46,XY,1qh+ - 758.580
46,XY,add(20)(p16qh+) - 758.530
46,XY,add(8)(p23) - 758.530
```

```
46,XY,del(13p) - 758.380
46,XY,del(15)(q11.2;q13) - 758.380
46,XY,del(22)(q11.2).ISH del(22)(q11.2;q11.2)(D22S7S-) - 758.380
46,XY,del(3)(q21;q23) - 758.380
46,XY,del(4)(p15.2) - 758.320
46.XY.del(5)(p14.1) - 758.310
46,XY,del(6)(q25.1;q25.31) - 758.380
46,XY,del(p13) - 758.310
46,XY,der(13)+(13:?)(q32:?) - 758.530
46,XY,der(13)+(13:?)(q32:?) - 758.540
46,XY,der(13:13)(q10;q10),+13 - 758.120
46,XY,der(13;14)(q10;q10)+14/45,XY,der(13;14)(q10;q10) - 758.520
46,XY,der(14:21)(q10;q10),+21 - 758.020
46,XY,der(14;21)(q10,q10) - 758.020
46,XY,der(21;21)(q10;q10),+21 - 758.020
46,XY,der(4)(p15.3).ISH DER (4) (WCP4-, D4F26-, D4596-) - 758.320
46,XY female, with a diagnosis of androgen insensitivity - 257.800
46,XY female, without a diagnosis of androgen insensitivity - 758.880
46,XY,inv(1)(p32;q31),3+der(3)+(1;3)(q31;p24) - 758.400
46,XY,inv(12) - 758.580
46,XY,inv(9)(p11;q12) - 758.580
46,XY,inv(9)(p12;q13) - 758.580
46,XY,inv(9)(p12;q13)mat,17 CHEV,+pat - 758.580
46,XY,inv(9)(pg12a13) - 758.580
46,XY,ISH del(22)(q11.2;q11.2)(D22S75-) - 758.380
46,XY,rob(14q;21q) - 758.540
46,XY,rob(14q;21q) - 758.020
46,XY,t(14q;21q) - 758.020
46,XY,t(16:17)(q13:q23) - 758.540
46,XY,t(17;19)(q21.2;q13.2) - 758.400
46,XY,t(3;18)(p13;q23) - 758.400
46,XY,t(4;14) - 758.400
46,XY,var(15)(q11.2) - 758.580
46,XY,var(15q) - 758.580
46,XY,var(22) - 758.580
46,XY,var21(+p) - 758.580
46,XY.ISH del(22)(q11.2;q11.2)(D22S75-) - 758.380
46,XY.ISH del(22)(q11.2;q11.2)(D22S75-) - 758.380
46,XY.ISH del(22)(q11.2)(D22S75X2) - 758.380
46,XY/45,X - 758.800
46,XY/45,XY,-14-18,+der(14)+(14;18) (q11.1;p11.2) - 758.540
46,XY/45,XY,-14-18,+der(14)+(14;18)(q11.1;p11.2) - 758.380
46,XY/45,XY,-19 - 758.380
46,XY/46,XY,-20,+der(20) - 758.580
46,XY/47,XXY (without Klinefelter syndrome phenotype) - 758.820
46,XY/47,XY+mar - 758.580
46,XY/47,XY,+16 - 758.520
46,XY/47,XY,+18 - 758.200
46,XY/47,XY,+21 - 758.040
46.XY/49.XXXXY (without Klinefelter syndrome phenotype) - 758.830
46, Y, der(X) - 758.880
47,X,fra(X)(q27.3)/47,XX,+21 - 758.000
47,XX,+(15;17)(q11.2?;q25),+18 - 758.200
47,XX,+1 - 758.520
47,XX,+10 - 758.510
47,XX,+11 - 758.510
47,XX,+12 - 758.510
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47,XX,+13 - 758.100
47,XX,+13,inv(9)(pg12;q13) - 758.100
47,XX,+13,inv(9)(pg12;q13) - 758.580
47,XX,+14 - 758.520
47,XX,+15 - 758.520
47.XX.+16 - 758.520
47,XX,+17 - 758.520
47,XX,+18 - 758.200
47,XX,+19 - 758.520
47,XX,+2 - 758.520
47,XX,+20 - 758.520
47,XX,+20(p10)/46,XX - 758.520
47,XX,+21 - 758.000
47,XX,+21,16(qh+) - 758.000
47,XX,+21,16qht - 758.000
47,XX,+21/46,XX - 758.040
47,XX,+21;inv(9)(p11;q12) - 758.000
47,XX,+22 - 758.520
47,XX,+3 - 758.520
47,XX,+4 - 758.520
47,XX,+5 - 758.520
47,XX,+6 - 758.510
47,XX,+7 - 758.510
47,XX,+8 - 758.500
47,XX,+8/46,XX - 758.500
47,XX,+9 - 758.510
47,XX,9qht,+21 - 758.000
47,XX,inv(9)(p11;q13),+21/46,XX,inv(9)(p11;q13) - 758.040
47,XX,inv(9)(p11;q13),+21/46,XX,inv(9)(p11;q13) - 758.580
47,XX,t(7;8)(q11.2;p21.1)+21 - 758.540
47,XX,t(7;8)(q11.2;p21.1),+21 - 758.000
47,XXX - 758.850
47,XXY - 758.700
47,XXY/46,XY - 758.820
47,XY,+1 - 758.520
47,XY,+10 - 758.510
47,XY,+11 - 758.510
47,XY,+12 - 758.510
47,XY,+13 - 758.100
47,XY,+14 - 758.520
47,XY,+15 - 758.520
47,XY,+16 - 758.520
47,XY,+17 - 758.520
47,XY,+18 - 758.200
47,XY,+18,inv(9)(p11;q12) - 758.200
47,XY,+19 - 758.520
47,XY,+2 - 758.520
47,XY,+20 - 758.520
47,XY,+21 - 758.000
47,XY,+21,+22 PSS - 758.000
47,XY,+21,1qht - 758.000
47,XY,+21/46,XY - 758.040
47.XY.+22 - 758.520
47,XY,+8/46,XY - 758.500
47,XY,+3 - 758.520
47,XY,+4 - 758.520
47,XY,+5 - 758.520
```

47,XY,+6 - 758.510 47,XY,+7 - 758.510 47,XY,+8 - 758.500 47,XY,+9 - 758.510 47,XY,+del(18)(q21.2) - 758.200 47,XY,+der(22) - 758.530 47,XY,+der(22)t(11,22)(q23;q11)mat - 758.530 47,XY,+mar - 758.580 47,XY,i(21)(q10)+mar - 758.580 47,XY,i(21)(q10)+mar - 758.020 47,XY,inv(2)(p11.2;q13),+21 - 758.000 47,XY,inv(9)(p11;q12),+21 - 758.000 47,XY,inv(9)(p11;q12),+21 - 758.000 47,XY,t(2;9)(p25.1;q34.11),+21 - 758.020 47,XYY - 758.840 47,XYY/46,XY - 758.840 48,XXXY - 758.710 48,XXYY - 758.710 48,XY,+21,+mar(pat) - 758.000 48,XY,+21,+mar(pat) - 758.580 49,XXXXY - 758.710 69,XXX - 758.586

69,XXY - 758.586

Appendix 5.2 6-Digit CDC Codes

BIRTH DEFECTS AND GENETIC DISEASES BRANCH 6-DIGIT CODE

For Reportable Congenital Anomalies

Based on the 1979 British Pediatric Association (BPA) Classification of Diseases and the World Health Organization's 1979 International Classification of Diseases, 9th Revision, Clinical Modification (ICD-9-CM)

Code modifications developed by Division of Birth Defects and Developmental Disabilities, National Center on Birth Defects and Developmental Disabilities, Centers for Disease Control and Prevention, Public Health Service, U.S. Department of Health and Human Services, Atlanta, Georgia 30333

Doc. No. 6digit88 Version 06/07 Replaces Versions 06/04 and 05/07

Explanation of 6-Digit Code

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6th Digit Code - Master

.000 Blank
.001 Left Only
.002 Right Only
.003 Unilateral Unspecified
.004 Bilateral
.005
.006
.007
.008 Possible, Probable, Borderline, or Rule Out;
Defects only diagnosed prenatally should be coded with the last digit 8 when the prenatal diagnosis is not definitive.
.009 Not Otherwise Specified (NOS)
```

Notes:

An asterisk (*) beside a disease code indicates that the code was created by CDC.

A pound symbol (#) beside a disease code indicates that the condition or defect is listed on the MACDP Exclusion List.

A check (T) beside a disease code indicates that an addition/revision was made since the last printing of the Procedure Manual. Use of the code should be according to the exclusion list criteria.

The abbreviations NEC and NOS used in this code are defined as not elsewhere classified and as not otherwise specified, respectively.

CONGENITAL ANOMALIES

Anencephalus and Similar Anomalies

740.0 Anencephalus

740.000 Absence of brain 740.010 Acrania 740.020 Anencephaly 740.030 Hemianencephaly, hemicephaly 740.080 Other

740.1 Craniorachischisis

740.100 Craniorachischisis

740.2 Iniencephaly

- 740.200 Closed iniencephaly 740.210 Open iniencephaly 740.290 Unspecified iniencephaly
- 741 Spina Bifida

741.0 Spina Bifida with Hydrocephalus

741.000 Spina bifida aperta, any site, with hydrocephalus 741.010 Spina bifida cystica, any site, with hydrocephalus and Arnold-Chiari malformation Arnold-Chiari malformation, NOS 741.020 Spina bifida cystica, any site, with stenosed aqueduct of Sylvius 741.030 Spina bifida cystica, cervical, with unspecified hydrocephalus Spina bifida cystica, cervical, with hydrocephalus but without mention of Arnold-Chiari malformation or aqueduct stenosis 741.040 Spina bifida cystica, thoracic, with unspecified hydrocephalus, no mention of Arnold-Chiari 741.050 Spina bifida cystica, lumbar, with unspecified hydrocephalus, no mention of Arnold-Chiari 741.060 Spina bifida cystica, sacral, with unspecified hydrocephalus, no mention of Arnold-Chiari 741.070 Spina bifida of any site with hydrocephalus of late onset

- 741.080 Other spina bifida, meningocele of specified site with hydrocephalus 741.085 Spina bifida, meningocele, cervicothoracic, with hydrocephalus 741.086 Spina bifida, meningocele thoracolumbar, with
- hydrocephalus
- 741.087 Spina bifida, meningocele, lumbosacral with hydrocephalus
- 741.090 Spina bifida of any unspecified type with hydrocephalus

741.9 Spina bifida without mention of hydrocephalus

- 741.900 Spina bifida (aperta), without hydrocephalus
- 741.910 Spina bifida (cystica), cervical, without hydrocephalus 741.920 Spina bifida (cystica), thoracic, without hydrocephalus 741.930 Spina bifida (cystica), lumbar, without hydrocephalus
- 741.940 Spina bifida (cystica), sacral, without hydrocephalus
- 741.980 Spina bifida, other specified site, without hydrocephalus Includes: cervicothoracic, thoracolumbar, lumbosacral
- 741.985 Lipomyelomeningocele
- 741.990 Spina bifida, site unspecified, without hydrocephalus (myelocoele, myelomeningocele, meningomyelocele)

742 Other Congenital Anomalies of Nervous System

742.0 Encephalocele

- 742.000 Occipital encephalocele
- 742.080 Other encephalocele of specified site (includes midline defects)
- 742.085 Frontal encephalocele
- 742.086 Parietal encephalocele
- 742.090 Unspecified encephalocele

742.1 Microcephalus

742.100 Microcephalus

742.2 Reduction deformities of brain

- 742.200 Anomalies of cerebrum
- 742.210 Anomalies of corpus callosum
- 742.220 Anomalies of hypothalamus
- 742.230 Anomalies of cerebellum
- 742.240 Agyria and lissencephaly
- 742.250 Microgyria, polymicrogyria
- 742.260 Holoprosencephaly
- 742.270 Arrhinencephaly
- 742.280 Other specified reduction defect of brain
- 742.290 Unspecified reduction defect of brain

742.3 Congenital hydrocephalus

Excludes: hydrocephalus with any condition in 741.9 (use 741.0)

- 742.300 Anomalies of aqueduct of Sylvius
- 742.310 Atresia of foramina of Magendie and Luschka

#	742.380	Includes: communicating hydrocephaly Hydrocephalus secondary to intraventricular hemorrhage (IVH) or CNS bleed	
742.4	Other specified anomalies of brain		
	742.400	Enlarged brain and/or head megalencephaly macrocephaly	
	742.410		
	742.420	Cerebral cysts	
		Other specified anomalies of brain Includes: cortical atrophy	
		cranial nerve defects	
	742.485	Ventricular cysts	
	742.486	Excludes: arachnoid cysts Small brain	
742.5	Other specified anomalies of spinal cord		
	742.500	Amuolia	
	742.500	Hypoplasia and dysplasia of spinal cord	
	, 12.010	atelomyelia	
		myelodysplasia	
	742.520	Diastematomyelia	
	742.530	Other cauda equina anomalies	
	742.540	Hydromyelia	
	540 500	Hydrorachis	
	742.580	Other specified anomalies of spinal cord and membranes Includes: congenital tethered cord	
742 0	Other and	ecified anomalies of nervous system	
742.0		congenital oculofacial paralysis	
	Excludes.	Moebius syndrome (use 352.600)	
	742 800	Jaw-winking syndrome	
	, 12,000	Marcus Gunn syndrome	
	742.810	Familial dysautonomia	
		Riley-Day syndrome	
	742.880	Other specified anomalies of nervous system	
742.9	Unspecifi	ed anomalies of brain, spinal cord and nervous systems	
	742.900	Brain, unspecified anomalies	
	742.910	Spinal cord, unspecified anomalies	
	742.990	Nervous system, unspecified anomalies	

743 Congenital Anomalies of Eye

743.000 Anophthalmos agenesis of eye cryptophthalmos
743.100 Microphthalmos, small eyes aplasia of eye hypoplasia of eye dysplasia of eye rudimentary eye

743.2 Buphthalmos

743.200 Buphthalmos congenital glaucoma hydrophthalmos
743.210 Enlarged eye, NOS
743.220 Enlarged cornea keratoglobus congenital megalocornea

743.3 Congenital cataract and lens anomalies

743.400 Corneal opacity

743.300 Absence of lens
congenital aphakia

743.310 Spherical lens
Spherophakia

743.320 Cataract, NOS

743.325 Cataract, anterior polar

743.326 Cataract, other specified

743.330 Displaced lens

743.340 Coloboma of lens

743.380 Other specified lens anomalies

743.390 Unspecified lens anomalies

743.4 Coloboma and other anomalies of anterior segments

743.410 Other corneal anomalies Excludes: megalocornea (use 743.220) 743.420 Absence of iris aniridia 743.430 Coloboma of iris 743.440 Other anomalies of iris polycoria ectopic pupil Peter's anomaly Excludes: brushfield spots (use 743.800) 743.450 Blue sclera If <36 weeks gestation, code only if another reportable defect is present. Always code if ≥36 weeks gestation. 743.480 Other specified colobomas and anomalies of anterior segments Rieger's anomaly 743.490 Unspecified colobomas and anomalies of anterior eye segments

743.5 Congenital anomalies of posterior segment

743.500 Specified anomalies of vitreous humour
743.510 Specified anomalies of retina
congenital retinal aneurysm
Excludes: Stickler syndrome (use 759.860)

743.520 Specified anomalies of optic disc
hypoplastic optic nerve
coloboma of the optic disc

743.530 Specified anomalies of choroid
743.535 Coloboma of choroid
743.536 Other specified anomalies of posterior segment of eye
743.590 Unspecified anomalies of posterior segment of eye

743.6 Congenital anomalies of eyelids, lacrimal system, and orbit

743.600 Blepharoptosis congenital ptosis 743.610 Ectropion 743.620 Entropion # 743.630 Other anomalies of eyelids absence of eyelashes long eyelashes weakness of eyelids Т fused eyelids (exclude if <25 weeks gestation unless another reportable defect is present) 743.635 Blepharophimosis small or narrow palpebral fissures 743.636 Coloboma of the eyelids 743.640 Absence or agenesis of lacrimal apparatus absence of punctum lacrimale 743.650 Stenosis or stricture of lacrimal duct 743.660 Other anomalies of lacrimal apparatus (e.g., cyst) 743.670 Anomalies of orbit

743.8 Other specified anomalies of eye

- * 743.810 Epibulbar dermoid cyst

743.9 Unspecified anomalies of eye

743.900 Unspecified anomalies of eye congenital: of eye (any part) anomaly, NOS deformity, NOS

744 Congenital Anomalies of Ear, Face, and Neck

744.0 Anomalies of ear causing impairment of hearing

- 744.000 Absence or stricture of auditory canal 744.010 Absence of auricle (pinna) absence of ear, NOS 744.020 Anomaly of middle ear fusion of ossicles 744.030 Anomaly of inner ear Includes: congenital anomaly of membranous labyrinth organ of Corti
- 744.090 Unspecified anomalies of ear with hearing impairment Includes: congenital deafness, NOS

744.1 Accessory auricle

- # 744.100 Accessory auricle
 - Polyotia
- 744.110 Preauricular appendage, tag, or lobule (in front of ear canal)
- 744.120 Other appendage, tag, or lobule include papillomas, ear tags

744.2 Other specified anomalies of ear

- 744.200 Macrotia (enlarged pinna)
- 744.210 Microtia (hypoplastic pinna and absence or stricture of external auditory meatus)
- 744.220 Bat ear
- Τ # 744.230 Other misshapen ear

pointed ear

elfin

pixie-like

lop ear

cauliflower ear

cleft in ear

malformed ear

absent or decreased cartilage

- 744.240 Misplaced ears
- 744.245 Low set ears 744.246 Posteriorly rotated ears
 - 744.250 Absence or anomaly of eustachian tube
- 744.280 Other specified anomalies of ear (see also 744.230)
- Excludes: Darwin's tubercle

744.3 Unspecified anomalies of ear

744.300 Unspecified anomalies of ear Congenital: ear (any part) anomaly, deformity, NOS

744.4 Branchial cleft, cyst, or fistula; preauricular sinus

744.400 Branchial cleft, sinus, fistula cyst, or pit # 744.410 Preauricular sinus, cyst, or pit

744.480 Other branchial cleft anomalies Includes: dermal sinus of head

744.500 Webbing of neck

Includes: pterygium colli,

redundant neck skin folds

744.8 Other unspecified anomalies of face and neck

744.800 Macrostomia (large mouth) 744.810 Microstomia (small mouth)

744.830 Macrocheilia (large lips)
744.830 Microcheilia (small lips)
744.880 Other specified anomalies of face/neck

744.9 Unspecified anomalies of face and neck

744.900 Congenital anomaly of neck, NOS Includes: short neck

744.910 Congenital anomaly of face, NOS Abnormal facies

745 Bulbus Cordis Anomalies and Anomalies of Cardiac Septal Closure

745.0 Common truncus (see 747.200 for pseudotruncus)

```
745.000 Persistent truncus arteriosus
absent septum between aorta and pulmonary
artery
745.010 Aortic septal defect
Includes: aortopulmonary window
```

745.1 Transposition of great vessels

```
745.100 Transposition of great vessels, complete (no VSD)
       745.110 Transposition of great vessels, incomplete (w/ VSD)
               Taussig-Bing syndrome
       745.120 Corrected transposition of great vessels,
               L-transposition, ventri in version
               Excludes: dextrocardia (use 746.800)
      745.130 Double outlet right ventricle (DORV) with normally
               related great vessels
      745.140 Double outlet right ventricle (DORV) with transposed
               great vessels
      745.150 Double outlet right ventricle (DORV), relationship of great
N
               vessels not specified
      745.180 Other specified transposition of great vessels,
N
               no mention of double outlet right ventricle (DORV)
      745.190 Unspecified transposition of great vessels
```

Excludes: atrial septal defect (use 745.590)

745.2 Tetralogy of Fallot

```
745.200 Fallot's tetralogy
745.210 Fallot's pentalogy
Fallot's tetralogy plus ASD
```

745.3 Single ventricle

745.300 Single ventricle
Common ventricle
Cor triloculare biatriatum

745.4 Ventricular septal defect

```
745.400 Roger's disease
               Note: This is an oudated term and the code is no longer
                      used. If this diagnostic term is encountered in
                      the medical record, code it as a ventricular septal
                      defect.
      745.410 Eisenmenger's syndrome
      745.420 Gerbode defect
Т
      745.480 Other specified ventricular septal defect
               Includes: cystalline
                           sub-cystalline
                           subarterial
                           conoventricular
      745.485
              Perimembranous VSD
               Includes: membranous VSD
```

```
745.486
        Muscular VSD
         Includes: mid-muscular and apical VSDs
745.487
         Inlet VSD
         Includes: common atrioventricular (AV) canal type VSD
         Note: Code common atrioventricular (AV) canal as
                745,630
                Code common atrioventricular (AV) canal with
                muscular VSD as 745.620
745.490
         Ventricular septal defect, NOS
         Excludes: common atrioventricular canal type (use
         745.620)
745.498
        Probable VSD
```

745.5 Ostium secundum type atrial septal defect

N # 745.500 Nonclosure of foramen ovale, NOS Patent foramen ovale (PFO) 1)Always code if ≥36 weeks of gestation at birth and defect last noted at ≥ 6 weeks of age. 2)If ≥36 weeks gestation at birth and defect last noted <6 weeks of age, code only if another reportable heart defect is present. 3) Never code if <36 weeks gestation at birth regardless of presence of other defects. 745.510 Ostium (septum) secundum defect Ν 745.520 Lutembacher's syndrome Note: This is an outdated term and the code is no longer used. If this diagnostic term is encountered in the medical record, code the individual components, not the syndrome. 745.580 Other specified atrial septal defect 745.590 ASD (atrial septal defect), NOS Auricular septal defect, NOS Partial foramen ovale PFO vs. ASD

745.6 Endocardial cushion defects

745.600 Ostium primum defects
745.610 Single common atrium, cor triloculare biventriculare
N 745.620 Common atrioventricular canal with ventricular septal defect (VSD)
Includes: Common AV canal with muscular VSD
Excludes: Inlet VSD or common AV canal type VSD (code as 745.487)
745.630 Common atrioventricular canal
745.680 Other specified cushion defect
745.690 Endocardial cushion defect, NOS

745.7 Cor biloculare

745.700 Cor biloculare

745.8 Other specified defects of septal closure

745.800 Other specified defects of septal closure

745.9 Unspecified defect of septal closure

745.900 Unspecified defect of septal closure

746 Other Congenital Anomalies of Heart

746.0 Anomalies of pulmonary valve

746.000 Atresia, hypoplasia of pulmonary valve Note: Code pulmonary artery atresia as 747.300 Code pulmonary artery hypoplasia as 747.380 Code "pulmonic" or "pulmonary" atresia or hypoplasia, NOS (no mention of valve or artery) as 746.995 Ν 746.010 Stenosis of pulmonary valve Excludes: pulmonary infundibular stenosis (use 746.830) Note: Code pulmonary artery stenosis as 747.320 Code "pulmonic" or "pulmonary" stenosis, NOS (no mention of valve or artery) as 746.995 N Pulmonary valve insufficiency or regurgitation, # 746.020 congenital Never code cases designated as 'mild', minimal', 'trivial', or 'physiologic'. Code cases designated as 'moderate' or 'severe' and those where the degree is not specified (NOS) only if another reportable heart defect is present. Other specified anomalies of pulmonary valve 746.080 pulmonary infundibular Excludes: stenosis (use 746.830) 746.090 Unspecified anomaly of pulmonary valve

746.1 Tricuspid atresia and stenosis

N 746.100 Tricuspid atresia only
Excludes: tricuspid stenosis and hypoplasia

N # 746.105 Tricuspid valve insufficiency or regurgitation,
congenital

Never code cases designated as 'mild', minimal', 'trivial',
or 'physiologic'.
Code cases designated as 'moderate' or 'severe' and those
where the degree is not specified (NOS) only if another
reportable heart defect is present.

746.106 Tricuspid stenosis or hypoplasia

746.2 Ebstein's anomaly

746.200 Ebstein's anomaly

746.3 Congenital stenosis of aortic valve

746.300 Congenital stenosis of aortic valve
Includes: congenital aortic stenosis
subvalvular aortic stenosis
Excludes: supravalvular aortic stenosis (747.220)

746.4 Congenital insufficiency of aortic valve

- N # 746.400 Aortic valve insufficiency or regurgitation, congenital
 Excludes: bicuspid aortic valve.
 Never code cases designated as 'mild', minimal', 'trivial',
 or 'physiologic'.
 Code cases designated as 'moderate' or 'severe' and those
 where the degree is not specified (NOS) only if another
 reportable heart defect is present.

 N 746.470 Bicuspid aortic valve
 * 746.480 Other specified anomalies of the aortic valves
 Includes: aortic valve atresia
 Excludes: supravalvular aortic stenosis (747.220)
- 746.5 Congenital mitral stenosis
 - 746.500 Congenital mitral stenosis 746.505 Absence, atresia, or hypoplasia of mitral valve

746.6 Mitral valve insufficiency or regurgitation, congenital

* 746.490 Unspecified anomalies of the aortic valves

N # 746.600 Mitral valve insufficiency or regurgitation, congenital Never code cases designated as 'mild', minimal', 'trivial', or 'physiologic'. Code cases designated as 'moderate' or 'severe' and those where the degree is not specified (NOS) only if another reportable heart defect is present.

746.7 Hypoplastic left heart syndrome

746.700 Hypoplastic left heart syndrome
Atresia, or marked hypoplasia of the
ascending aorta and defective development
of left ventricle (with mitral valve atresia)

746.8 Other specified anomalies of the heart

746.800 Dextrocardia without with no mention of situs inversus Excludes: dextrocardia with situs inversus use 759.300)

746.810 Levocardia

Note: This condition has been moved to the never code list.

746.820 Cor triatriatum

746.830 Pulmonary infundibular (subvalvular) stenosis

746.840 Trilogy of Fallot

746.850 Anomalies of pericardium

746.860 Anomalies of myocardium cardiomegaly, congenital, NOS cardiomyopathy, congenital cardiomyopathy, hypertrophic

Note: Do not code cardiomyopathy of any type in a newborn of a diabetic mother (either gestational or preexisting diabetes).

```
746.870 Congenital heart block
      746.880 Other specified anomalies of heart
               Includes:
                             ectopia (ectopic) cordis (mesocardia),
                             conduction defects, NOS
      746.881 Hypoplastic left ventricle
               Excludes:
                            hypoplastic left heart syndrome (746.700)
      746.882 Hypoplastic right heart (ventricle)
               Uhl's disease
     746.883 Hypoplastic ventricle, NOS
      746.885 Anomalies of coronary artery or sinus
      746.886 Ventricular hypertrophy (right or left)
Ν
               Note: Do not code ventricular hypertrophy of any type in
               a newborn of a diabetic mother (either gestational or
               pre-existing diabetes).
      746.887 Other defects of the atria
                             congenital Wolfe-Parkinson-White
               Excludes:
                             (use 426.705)
```

rhythm anomalies (use 426.-, 427.-)

746.9 Unspecified anomalies of heart

	746.900	Unspecified anomalies of heart valves
	746.910	Anomalous bands of heart
	746.920	Acyanotic congenital heart disease, NOS
	746.930	Cyanotic congenital heart disease, NOS
		Blue baby
	746.990	Unspecified anomaly of heart:
		Includes: congenital heart disease (CHD)
N	746.995	"Pulmonic" or "pulmonary" atresia, stenosis, or
		hypoplasia, NOS (no mention of valve or artery)
		Note: Code pulmonary valve atresia or hypoplasia as
		746.000
		Code pulmonary valve stenosis as 746.010
		Code pulmonary artery atresia as 747.300
		Code pulmonary artery stenosis as 747.320
		Code pulmonary artery hypoplasia as 747.380

747 Other Congenital Anomalies of Circulatory System

N # 747.000 Patent ductus arteriosus (PDA)
Note: 1)Always code if ≥36 weeks of gestation at birth and
defect last noted at ≥6 weeks of age.
2)If ≥36 weeks gestation at birth and defect last noted
<6 weeks of age, code only if the PDA was treated (e.g.
by ligation or indomethicin) or if another reportable
heart defect is present.
3)Never code if <36 weeks gestation at birth or if
treated with prostaglandins regardless of gestational
age. (See PDA Tree Appendix)
747.008 Probable PDA</pre>

747.1 Coarctation of aorta

- 747.100 Preductal (proximal) coarctation of aorta 747.110 Postductal (distal) coarctation of aorta 747.190 Unspecified coarctation of aorta
- R = Rev. 6/07 N = Rev. 5/07 T = Rev. 6/04* = code created by CDC
 # = on the MACDP Excl List

747.2 Other anomalies of aorta

```
747.200 Atresia of aorta
               absence of aorta
               pseudotruncus arteriosus
      747.210 Hypoplasia of aorta
               tubular hypoplasia of aorta
      747.215 Interrupted aortic arch, Type A
      747.216 Interrupted aortic arch, Type B
Ν
      747.217 Interrupted aortic arch, Type C
Ν
      747.220 Supra-aortic stenosis (supravalvular)
               Excludes: aortic stenosis,
                         congenital (see 746.300)
      747.230 Persistent right aortic arch
      747.240 Aneurysm of sinus of Valsalva
               Vascular ring (aorta)
      747.250
               double aortic arch
                Includes:
                             vascular ring compression of trachea
      747.260
               Overriding aorta
               dextroposition of aorta
       747.270 Congenital aneurysm of aorta
               congenital dilatation of aorta
      747.280
               Other specified anomalies of aorta
       747.285 Interrupted aortic arch, NOS, type not specified
Ν
       747.290 Unspecified anomalies of aorta
```

747.3 Anomalies of pulmonary artery

```
747.300
                Pulmonary artery atresia, absence or agenesis
Ν
                Note: Code pulmonary valve atresia as 746.000
                       Code "pulmonic" or "pulmonary" atresia, NOS (no
                       mention of valve or artery) as 746.995
      747.310 Pulmonary artery atresia with septal defect
      747.320
M
               Pulmonary artery stenosis
                Includes: Stenosis of the main pulmonary artery or of
                          the right or left main branches
                Note: Code pulmonary valve stenosis as 746.010
                       Code "pulmonic" or "pulmonary" stenosis, NOS (no
                       mention of valve or artery) as 746.995
N
      747.325
               Peripheral pulmonary artery stenosis
                Includes: Stenosis of a pulmonary artery peripheral to
                          the main right or left main branches
                          Peripheral pulmonic stenosis (PPS), NOS,
                          documented by echocardiogram
                Excludes: Peripheral pulmonic stenosis (PPS) murmur only
                          (not documented by echocardiogram)
                Note: 1) Always code if ≥36 weeks of gestation at birth and
                defect last noted at ≥6 weeks of age.
                2) If ≥36 weeks gestation at birth and defect last noted
                <6 weeks of age, code only if another reportable heart
                defect is present.
                3) Never code if <36 weeks gestation at birth.
                (See PPS Tree Appendix)
      747.330
               Aneurysm of pulmonary artery
                dilatation of pulmonary artery
      747.340 Pulmonary arteriovenous malformation or aneurysm
```

```
747.380 Other specified anomaly of pulmonary artery Includes: pulmonary artery hypoplasia
Note: Code pulmonary valve hypoplasia as 746.000
Code "pulmonic" or "pulmonary" hypoplasia, NOS
(no mention of valve or artery) as 746.995
```

747.4 Anomalies of great veins

747.400 Stenosis of vena cava (inferior or superior)
747.410 Persistent left superior vena cava
747.420 (TAPVR) Total anomalous pulmonary venous return
747.430 Partial anomalous pulmonary venous return
747.440 Anomalous portal vein termination
747.450 Portal vein - hepatic artery fistula
747.480 Other specified anomalies of great veins

747.390 Unspecified anomaly of pulmonary artery

747.490 Unspecified anomalies of great veins

747.5 Absence or hypoplasia of umbilical artery

747.500 Single umbilical artery

747.6 Other anomalies of peripheral vascular system

747.600 Stenosis of renal artery 747.610 Other anomalies of renal artery 747.620 Arteriovenous malformation (peripheral) Excludes: pulmonary (747.340) cerebral (747.800) retinal (743.510) 747.630 Congenital phlebectasia congenital varix 747.640 Other anomalies of peripheral arteries Includes: aberrant subclavian artery 747.650 Other anomalies of peripheral veins Budd-Chiari - occlusion of hepatic vein (use Excludes: 453.000) 747.680 Other anomalies of peripheral vascular system Includes: primary pulmonary artery hypertension ONLY if it is present in an infant at >7 days of age 747.690 Unspecified anomalies of peripheral vascular system

747.8 Other specified anomalies of circulatory system

747.9 Unspecified anomalies of circulatory system

747.900 Unspecified anomalies of circulatory system

748 Congenital Anomalies of Respiratory System

748.0 Choanal atresia

748.000 Choanal atresia atresia of nares, anterior or posterior congenital stenosis

748.1 Other anomalies of nose

748.100 Agenesis or underdevelopment of nose
748.110 Accessory nose
748.120 Fissured, notched, or cleft nose
748.130 Sinus wall anomalies
748.140 Perforated nasal septum

748.180 Other specified anomalies of nose
flat bridge of nose
wide nasal bridge
small nose and nostril
absent nasal septum

748.185 Tubular nose, single nostril, proboscis
748.190 Unspecified anomalies of nose
Excludes: congenital deviation of the nasal
septum (use 754.020)

748.2 Web of larynx

748.205 Web of larynx-glottic 748.206 Web of larynx-subglottic 748.209 Web of larynx-NOS

748.3 Other anomalies of larynx, trachea, and bronchus

- 748.300 Anomalies of larynx and supporting cartilage

 748.310 Congenital subglottic stenosis Never code if chart states the condition was acquired or secondary to endotracheal (ET) intubation or ventilation

 748.330 Other anomalies of trachea

 Excludes: vascular ring compression of the trachea (use 747.250)

 748.340 Stenosis of bronchus

 748.350 Other anomalies of bronchus

 748.360 Congenital laryngeal stridor, NOS
 - 748.380 Other specified anomalies of larynx and bronchus
 - 748.385 Cleft larynx, laryngotracheoesophageal cleft
 - 748.390 Unspecified anomalies of larynx, trachea, and bronchus

748.4 Congenital cystic lung

748.400 Single cyst, lung or lung cyst
748.410 Multiple cysts, lung
Polycystic lung
748.420 Honeycomb lung
748.480 Other specified congenital cystic lung

748.5 Agenesis or aplasia of lung

748.500 Agenesis or aplasia of lung

T 748.510 Hypoplasia of lung; Pulmonary hypoplasia

Exclude if isolated defect in infants <36 weeks gestation.

748.520 Sequestration of lung

748.580 Other specified dysplasia of lung

Fusion of lobes of lung

* 748.590 Unspecified dysplasia of lung

748.6 Other anomalies of lung

748.600 Ectopic tissues in lung
748.610 Bronchiectasis
748.620 Accessory lobe of lung
748.625 Bilobar right lung or right lung with left lung bronchial pattern
748.690 Other and unspecified anomalies of lung

748.8 Other specified anomalies of respiratory system

748.800 Anomaly of pleura
748.810 Congenital cyst of mediastinum
748.880 Other specified respiratory system anomalies
Includes: congenital lobar emphysema
lymphangiectasia of lungs

748.9 Unspecified anomalies of respiratory system

748.900 Unspecified anomalies of respiratory system
Absence of respiratory organ, NOS
Anomaly of respiratory system, NOS

749 Cleft Palate and Cleft Lip

749.0 Cleft palate alone

(If description of condition includes Pierre Robin sequence, use additional code, 524.080)

```
749.000 Cleft hard palate, unilateral
749.010 Cleft hard palate, bilateral
749.020 Cleft hard palate, central
749.030 Cleft hard palate, NOS
749.040 Cleft soft palate, alone unilateral
749.050 Cleft soft palate, alone bilateral
749.060 Cleft soft palate, alone central
749.070 Cleft soft palate, alone, NOS
749.080 Cleft uvula
749.090 Cleft palate, NOS
palatoschisis
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749.1 Cleft lip alone

749.2 Cleft lip with cleft palate

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749.200 Cleft lip, unilateral, with any cleft palate 749.210 Cleft lip, bilateral, with any cleft palate 749.220 Cleft lip, central, with any cleft palate 749.290 Cleft lip, NOS, with any cleft palate
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750 Other Congenital Anomalies of Upper Alimentary Tract

750.000 Tonque tie Ankyloglossia

750.1 Other anomalies of tongue

Excludes:protruding tongue (never a defect)

750.100 Aglossia Absence of tongue 750.110 Hypoglossia (small tongue) Microglossia 750.120 Macroglossia (large tongue) 750.130 Dislocation or displacement of tongue

Glossoptosis

750.140 Cleft tongue or split tongue

750.180 Other specified anomalies of tongue 750.190 Unspecified anomalies of tongue

750.2 Other specified anomalies of mouth and pharynx

750.200 Pharyngeal pouch

750.210 Other pharyngeal anomalies

750.230 Other anomalies of salivary glands or ducts

750.240 High arched palate

750.250 Other anomalies of palate

750.260 Lip fistulae or pits

750.270 Other lip anomalies

Includes: notched lip, prominent philtrum,

long philtrum

cleft lip (see 749) Excludes:

750.280 Other specified anomalies of mouth and pharynx

Excludes: receding jaw (see 524.0)

large and small mouth (see 744.8)

750.3 Tracheoesophageal (T-E) fistula, esophageal atresia and stenosis

750.300 Esophageal atresia without mention of T-E fistula

750.310 Esophageal atresia with mention of T-E fistula

750.320 Tracheoesophageal fistula without mention of esophageal atresia

750.325 Tracheoesophageal fistula - "H" type

750.330 Bronchoesophageal fistula with or without mention of esophageal atresia

750.340 Stenosis or stricture of esophagus

750.350 Esophageal web

750.380 Other tracheoesophageal anomalies

750.4 Other specified anomalies of esophagus

750.400 Congenital dilatation of esophagus

giant esophagus

750.410 Displacement of esophagus

750.420 Diverticulum of esophagus

esophageal pouch

750.430 Duplication of esophagus

750.480 Other specified anomalies of esophagus

750.5 Congenital hypertrophic pyloric stenosis

750.500 Pylorospasm
750.510 Congenital hypertrophic pyloric stenosis
750.580 Other congenital pyloric obstruction

750.6 Congenital hiatus hernia

750.600 Congenital hiatus hernia
Cardia displacement through esophageal hiatus
Partial thoracic stomach
Excludes: congenital diaphragmatic hernia (756.610)

750.7 Other specified anomalies of stomach

750.700 Microgastria
750.710 Megalogastria
750.720 Cardiospasm
achalasia of cardia, congenital
750.730 Displacement or transposition of stomach
750.740 Diverticulum of stomach
750.750 Duplication of stomach
750.780 Other specified anomalies of stomach

750.8 Other specified anomalies of upper alimentary tract

750.800 Other specified anomalies of upper alimentary tract

750.9 Unspecified anomalies of upper alimentary tract

750.900 Unspecified anomalies of mouth and pharynx 750.910 Unspecified anomalies of esophagus 750.920 Unspecified anomalies of stomach 750.990 Unspecified anomalies of upper alimentary tract

751 Other Congenital Anomalies of Digestive System

751.0 Meckel's diverticulum

- 751.000 Persistent omphalomesenteric duct persistent vitelline duct
- # 751.010 Meckel's diverticulum

751.1 Atresia and stenosis of small intestine

- 751.100 Stenosis, atresia or absence of duodenum
- 751.110 Stenosis, atresia or absence of jejunum
- 751.120 Stenosis, atresia or absence of ileum
- 751.190 Stenosis, atresia or absence of small intestine
- 751.195 Stenosis, atresia or absence of small intestine with fistula

751.2 Atresia and stenosis of large intestine, rectum and anal canal

- 751.200 Stenosis, atresia or absence of large intestine Stenosis, atresia or absence of appendix
- 751.210 Stenosis, atresia or absence of rectum with fistula
- 751.220 Stenosis, atresia or absence of rectum without mention of fistula
- 751.230 Stenosis, atresia or absence of anus with fistula Includes: imperforate anus with fistula
- 751.240 Stenosis, atresia or absence of anus without mention of fistula Includes: imperforate anus without fistula

751.3 Hirschsprung's disease and other congenital functional disorders of the colon

- 751.300 Total intestinal aganglionosis
- 751.310 Long-segment Hirschsprung's disease; aganglionosis beyond the rectum
- 751.320 Short-segment Hirschsprung's disease; aganglionosis involving no more than the anal sphincter and the rectum
- 751.330 Hirschsprung's disease, NOS
- 751.340 Congenital megacolon congenital macrocolon, not aganglionic

751.4 Anomalies of intestinal fixation

- 751.400 Malrotation of cecum and/or colon
- 751.410 Anomalies of mesentery
- 751.420 Congenital adhesions or bands of omentum and peritoneum; Ladd's bands
- 751.490 Other specified and unspecified malrotation
- 751.495 Malrotation of small intestine alone

751.5 Other anomalies of intestine

- 751.500 Duplication of anus, appendix, cecum, or intestine enterogenous cyst
- 751.510 Transposition of appendix, colon, or intestine
- 751.520 Microcolon

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751.530 Ectopic (displaced) anus
       751.540 Congenital anal fistula
       751.550 Persistent cloaca
R
       751.555 Exstrophy of cloaca
                Excludes exstrophy of urinary bladder not associated with
                imperforate anus (use 753.500)
    * 751.560 Duodenal web
    # 751.580 Other specified anomalies of intestine
                Includes: rectal fissures
       751.590 Unspecified anomalies of intestine
751.6 Anomalies of gallbladder, bile ducts, and liver
       751.600 Absence or agenesis of liver, total or partial
       751.610 Cystic or fibrocystic disease of liver
    # 751.620 Other anomalies of liver
                hepatomegaly
                hepatosplenomegaly (also use code 759.020)
                              Budd-Chiari (use 453.000)
                Excludes:
       751.630 Agenesis or hypoplasia of gallbladder
       751.640 Other anomalies of gallbladder
                duplication of gallbladder
       751.650 Agenesis or atresia of hepatic or bile ducts
                Includes:
                             biliary atresia
                Excludes:
                              congenital or neonatal hepatitis
                              (use 774.480 or 774.490)
      751.660 Choledochal cysts
751.670 Other anomalies of hepatic or bile ducts
751.680 Anomalies of biliary tract, NEC
751.7 Anomalies of pancreas
       Excludes: fibrocystic disease of pancreas (277.000)
                diabetes mellitus,
                congenital
                neonatal
       751.700 Absence, agenesis or hypoplasia of pancreas
       751.710 Accessory pancreas
       751.720 Annular pancreas
       751.730 Ectopic pancreas
       751.740 Pancreatic cyst
       751.780 Other specified anomalies of pancreas
       751.790 Unspecified anomalies of pancreas
751.8 Other specified anomalies of digestive system
       751.800 Absence of alimentary tract, NOS
                (complete or partial)
       751.810 Duplication of alimentary tract
       751.820 Ectopic digestive organs, NOS
       751.880 Other specified anomalies of digestive system
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751.9 Unspecified anomalies of digestive system

751.900 Unspecified anomalies of digestive system congenital of digestive system, NOS anomaly, NOS deformity, NOS obstruction, NOS

752 Congenital Anomalies of Genital Organs

Excludes: congenital hydrocele (778.600) testicular feminization syndrome (257.800) syndromes associated with anomalies in number and form of chromosomes (758)

752.0 Anomalies of ovaries

- 752.000 Absence or agenesis of ovaries
- 752.010 Streak ovary
- 752.010 Stream Ovary
- 752.080 Other specified anomalies of ovaries 752.085 Multiple ovarian cysts 752.090 Unspecified anomalies of ovaries

752.1 Anomalies of fallopian tubes and broad ligaments

- 752.100 Absence of fallopian tube or broad ligament
- 752.110 Cyst of mesenteric remnant epoophoron cyst

cyst of Gartner's duct

- 752.120 Fimbrial cyst
 - parovarian cyst
- 752.190 Other and unspecified anomalies of fallopian tube and broad ligaments

752.2 Doubling of uterus

752.200 Doubling of uterus doubling of uterus (any degree) or associated with doubling of cervix and vagina

752.3 Other anomalies of uterus

- 752.300 Absence or agenesis of uterus
- 752.310 Displaced uterus
- 752.320 Fistulae involving uterus with digestive or urinary tract

Includes: uterointestinal fistula uterovesical fistula

- 752.380 Other anomalies of uterus bicornuate uterus
 - unicornis uterus
- 752.390 Unspecified anomalies of uterus

752.4 Anomalies of cervix, vagina, and external female genitalia

- 752.400 Absence, atresia or agenesis of cervix
- 752.410 Absence or atresia of vagina, complete or partial
- 752.420 Congenital rectovaginal fistula
- 752.430 Imperforate hymen
- # 752.440 Absence or other anomaly of vulva

fusion of vulva

hypoplastic labia majora - Always code if $\geq 36 weeks$ gestation. If <36 weeks gestation, code only if another reportable defect is present.

752.450 Absence or other anomaly of clitoris Includes: clitoromegaly enlarged clitoris clitoral hypertrophy

prominent clitoris

Embryonal cyst of vagina # 752.460

752.470 Other cyst of vagina, vulva, or canal of Nuck

752.480 Other specified anomalies of cervix, vagina, or external female genitalia

> Includes: vaginal tags hymenal tags

752.490 Unspecified anomalies of cervix, vagina, or external female genitalia

752.5 Undescended testicle

1) If < 36 weeks gestation, code only if there is a medical/surgical intervention for this problem; 2) If ≥36 weeks gestation and defect last noted at <1 year of age, code only if there was a medical/surgical intervention for this problem or if another reportable defect is present 3)Always code if ≥36 weeks gestation and defect first noted at ≥ 1 of age.

752.500 Undescended testicle, unilateral undescended, unpalpable

752.501 Left undescended testicle

752.502 Right undescended testicle

752.514 Undescended testicle, bilateral

752.520 Undescended testicle, NOS (Cryptorchidism)

752.530 Ectopic testis, unilateral and bilateral

752.6 Hypospadias and epispadias

752.600 Hypospadias (alone), NOS

752.605 1°, glandular, coronal 752.606 2°, penile 752.607 3°, perineal, scrotal

752.610 Epispadias

752.620 Congenital chordee (with hypospadias), NOS

752.621 Congenital chordee alone (chordee w/o hypospadias)

752.625 Cong. chordee with 1° , coronal hypospadias

752.626 Cong. chordee with 2° , penile hypospadias

752.627 Cong. chordee with 3°, perineal, scrotal hypospadias

752.7 Indeterminate sex and pseudohermaphroditism

Excludes:pseudohermaphroditism:

female, with adrenocortical disorder (see 255.200) male, with gonadal disorder with specified chromosomal anomaly (see 758)

752.700 True hermaphroditism ovotestis

752.710 Pseudohermaphroditism, male
752.720 Pseudohermaphroditism, female
pure gonadal dysgenesis
Excludes: gonadal agenesis (758.690)
752.730 Pseudohermaphrodite, NOS
752.790 Indeterminate sex, NOS
ambiguous genitalia

752.8 Other specified anomalies of male genital organs

752.800 Absence of testis monorchidism, NOS # 752.810 Aplasia or hypoplasia of testis and scrotum 752.820 Other anomalies of testis and scrotum polyorchidism bifid scrotum Excludes: torsion of the testes or spermatic cord (use #608.200) 752.830 Atresia of vas deferens 752.840 Other anomalies of vas deferens and prostate 752.850 Absence or aplasia of penis 752.860 Other anomalies of penis absent or hooded foreskin redundant foreskin (never a defect) 752.865 Small penis, hypoplastic penis, or micropenis 752.870 Cysts of embryonic remnants cyst: hydatid of Morgagni Wolffian duct appendix testis 752.880 Other specified anomalies of genital organs microgenitalia macrogenitalia

752.9 Unspecified anomalies of genital organs

752.900 Unspecified anomalies of genital organs Congenital: of genital organ, NEC anomaly, NOS or deformity, NOS

753 Congenital Anomalies of Urinary System

753.0 Renal agenesis and dysgenesis

- 753.000 Bilateral absence, agenesis, dysplasia, or hypoplasia of kidneys Potter's syndrome 753.009 Renal agenesis, NOS
- 753.010 Unilateral absence, agenesis, dysplasia or hypoplasia of kidneys

753.1 Cystic kidney disease

- 753.100 Renal cyst (single)
- 753.110 Polycystic kidneys, infantile type
- 753.120 Polycystic kidneys, adult type
- 753.130 Polycystic kidneys, NOS
- 753.140 Medullary cystic disease, juvenile type
- 753.150 Medullary cystic disease, adult type
- Medullary sponge kidney
- 753.160 Multicystic renal dysplasia
- Multicystic kidney
- 753.180 Other specified cystic disease Includes: cystic kidneys, NOS

753.2 Obstructive defects of renal pelvis and ureter

- 753.200 Congenital hydronephrosis
- 753.210 Atresia, stricture, or stenosis of ureter

ureteropelvic junction obstruction/stenosis Includes: ureterovesical junction obstruction/stenosis hypoplastic ureter

- 753.220 Megaloureter, NOS
 - hydroureter Includes:
- 753.290 Other and unspecified obstructive defects of renal pelvis and ureter

753.3 Other specified anomalies of kidney

- 753.300 Accessory kidney
- 753.310 Double or triple kidney and pelvis

pyelon duplex or triplex

- 753.320 Lobulated, fused, or horseshoe kidney
- 753.330 Ectopic kidney
- 753.340 Enlarged, hyperplastic or giant kidney
- 753.350 Congenital renal calculi
- 753.380 Other specified anomalies of kidney

753.4 Other specified anomalies of ureter

- 753.400 Absence of ureter
- 753.410 Accessory ureter

double ureter, duplex collecting system

- 753.420 Ectopic ureter
- 753.480 Other specified anomalies of ureter Includes: ureterocele

753.5 Exstrophy of urinary bladder

753.500 Exstrophy of urinary bladder ectopia vesicae extroversion of bladder

753.6 Atresia and stenosis of urethra and bladder neck

- 753.600 Congenital posterior urethral valves or posterior urethral obstruction
- 753.610 Other atresia, or stenosis of bladder neck
- 753.620 Obstruction, atresia or stenosis of anterior urethra 753.630 Obstruction, atresia or stenosis of urinary meatus Includes: meatal stenosis
- 753.690 Other and unspecified atresia and stenosis of urethra and bladder neck

753.7 Anomalies of urachus

- T # 753.700 Patent urachus
 - 753.710 Cyst of urachus
 - 753.790 Other and unspecified anomaly of urachus

753.8 Other specified anomalies of bladder and urethra

- 753.800 Absence of bladder or urethra
- 753.810 Ectopic bladder
- 753.820 Congenital diverticulum or hernia of bladder
- 753.830 Congenital prolapse of bladder (mucosa)
- 753.840 Double urethra or urinary meatus
- 753.850 Ectopic urethra or urethral orifice
- 753.860 Congenital digestive-urinary tract fistulae rectovesical fistula
- 753.870 Urethral fistula, NOS
- 753.880 Other specified anomalies of bladder and urethra

753.9 Unspecified anomalies of urinary system

- 753.900 Unspecified anomaly of kidney
- 753.910 Unspecified anomaly of ureter
- 753.920 Unspecified anomaly of bladder
- 753.930 Unspecified anomaly of urethra
- 753.990 Unspecified anomaly of urinary system, NOS

754 Certain Congenital Musculoskeletal Anomalies

754.0 Of skull, face, and jaw Excludes: dentofacial anomalies (524.0) Pierre Robin sequence (524.080) syphilitic saddle nose (090.000) 754.000 Asymmetry of face 754.010 Compression (Potter's) facies # 754.020 Congenital deviation of nasal septum bent nose T 754.030 Dolichocephaly Always code if ≥36 weeks gestation # If <36 weeks gestation, code only if another reportable defect is present 754.040 Depressions in skull Includes: large fontanelle small fontanelle 754.050 Plagiocephaly 754.055 Asymmetric head * 754.060 Scaphocephaly, no mention of craniosynostosis * 754.070 Trigonocephaly, no mention of craniosynostosis Always code if ≥ 36 weeks gestation # If <36 weeks gestation, code only if another reportable defect is present * 754.080 Other specified skull deformity, no mention of craniosynostosis Includes:brachycephaly acrocephaly

754.1 Anomalies of sternocleidomastoid muscle

* 754.090

754.100 Anomalies of sternocleidomastoid muscle

* Includes: absent or hypoplastic sternocleidomastoid contracture of sternocleidomastoid muscle sternomastoid tumor

Excludes: congenital sternocleidomastoid torticollis (use 756.860)

754.2 Certain congenital musculoskeletal deformities of spine

turricephaly oxycephaly

Deformity of skull, NOS

754.200 Congenital postural scoliosis 754.210 Congenital postural lordosis 754.220 Congenital postural curvature of spine, NOS

754.3 Congenital dislocation of hip

754.300 Congenital dislocation of hip
754.310 Unstable hip
preluxation of hip
subluxation of hip
predislocation status of hip at birth

Τ

754.4 Congenital genu recurvatum and bowing of long bones of leg

- 754.400 Bowing, femur
- 754.410 Bowing, tibia and/or fibula
- 754.420 Bow legs, NOS
- 754.430 Genu recurvatum
- 754.440 Dislocation of knee, congenital
- 754.490 Deformity of leg, NOS

754.5 Varus (inward) deformities of feet

- 754.500 Talipes equinovarus 754.510 Talipes calcaneovarus
- # 754.520 Metatarsus varus or metatarsus adductus
 - 754.530 Complex varus deformities
 - 754.590 Unspecified varus deformities of feet

754.6 Valgus (outward) deformities of feet

- 754.600 Talipes calcaneovalgus
- 754.610 Congenital pes planus
- 754.615 Pes valgus
- 754.680 Other specified valgus deformities of foot
- 754.690 Unspecified valgus deformities of foot

754.7 Other deformities of feet

- 754.700 Pes cavus
 - Claw foot (use 755.350 for claw foot)
- 754.720 Short Achilles tendon
- 754.730 Clubfoot, NOS
 - talipes, NOS
- 754.735 Congenital deformities of foot, NOS
- 754.780 Other specified deformities of ankle and/or toes
 - Includes: dorsiflexion of foot
 - Excludes: widely spaced 1st and 2nd toes (use 755.600)

754.8 Other specified congenital musculoskeletal deformities

- 754.800 Pigeon chest (pectus carinatum)
- 754.810 Funnel chest (pectus excavatum)
- 754.820 Other anomalies of chest wall
 - Includes: deformed chest, barrel chest
- 754.825 Shield chest
- 754.830 Dislocation of elbow
- 754.840 Club hand or fingers
- 754.850 Spade-like hand
- 754.880 Other specified deformity of hands
 - (see 755.500 for specified anomalies of fingers)

755 Other Congenital Anomalies of Limbs

755.0 Polydactyly

- 755.005 Accessory fingers (postaxial polydactyly, Type A) # 755.006 Skin tag (postaxial polydactyly, Type B) Exclude: Do not code in black infants. 755.007 Unspecified finger or skin tag (postaxial polydactyly, NOS)
 755.010 Accessory thumbs (preaxial polydactyly)
 755.020 Accessory toes (postaxial)
 755.030 Accessory big toe (preaxial)
 755.090 Accessory digits, NOS (hand/foot not specified)
 755.095 Accessory digits hand, NOS (preaxial, postaxial not
 - specified)
 - 755.096 Accessory digits foot, NOS (preaxial, postaxial not specified)

755.1 Syndactyly

- 755.100 Fused fingers
- 755.110 Webbed fingers
- 755.120 Fused toes
- # 755.130 Webbed toes

Code webbing of the second and third toes only if another reportable defect is present. Always code webbing of other toes regardless of whether another reportable defect is present

- 755.190 Unspecified syndactyly (see below for specified site)
- 755.191 Unspecified syndactyly thumb and/or fingers, unilateral
- 755.192 Unspecified syndactyly thumb and/or fingers, bilateral
- 755.193 Unspecified (webbed vs. fused) syndactyly thumb and/or fingers, NOS
- 755.194 Unspecified syndactyly toes unilateral
- 755.195 Unspecified syndactyly toes bilateral
- 755.196 Unspecified syndactyly toes, NOS
- 755.199 Unspecified syndactyly (i.e., webbed vs. fused) digits not known

755.2 Reduction defects of upper limb

- Т If description of the condition includes amniotic or constricting bands use additional code, 658.800 (Only use 658.800 if another reportable defect is present) Excludes shortening of upper limb (use 755.580) or hypoplasia of upper limb (use 755.585)
 - 755.200 Absence of upper limb

humerus (total or partial), radius, ulna and hand Absent:

Includes: amelia of upper limb, NOS

infants with rudimentary or nubbin fingers

attached to stump of humerus or shoulder girdle

755.210 Absence of upper arm and forearm

humerus (total or partial), radius and ulna Absent:

(total or partial)

Present: hand (total or partial)

Includes: phocomelia of upper limb, NOS;

intercalary reduction defect of upper limb, NOS

755.220 Absence of forearm only or upper arm only Absent: radius and ulna Present: humerus, hand (total or partial) or Absent: humerus Present: radius, ulna, and hand 755.230 Absence of forearm and hand Absent: radius and ulna (total or partial) and hand Includes: infants with rudimentary or nubbin fingers attached to stump of forearm or elbow 755.240 Absence of hand or fingers Absent: hand or fingers (total or partial) not in conjunction with ray or long bone reduction Includes: rudimentary or nubbin fingers; absent individual phalanges; absent or missing fingers, NOS Excludes: isolated absent or hypoplastic thumb (use 755.260) 755.250 Split-hand malformation central fingers (third with or without second, Absent: fourth) and metacarpals (total or partial) Includes: monodactyly; lobster-claw hand Excludes: isolated absent central fingers without metacarpal defects (use 755.240) 755.260 Preaxial longitudinal reduction defect of upper limb radius (total or partial) and/or thumb with or Absent: without second finger (total or partial) Includes: isolated absent or hypoplastic thumb; radial ray defect, NOS 755.265 Longitudinal reduction defect of upper limb, NOS Includes: absent forearm long bone with absent fingers, 755.270 Postaxial longitudinal reduction defect of upper limb Includes: isolated absent ulna (total or partial); absent fifth with or without fourth finger (total or partial) only if ulna or fifth \pm fourth metacarpal also totally or partially absent;

755.3 Reduction defects of lower limb

T If description of condition includes amniotic or constricting bands use additional code, 658.800 (Only use this code if another reportable defect is present)

ulnar ray defect, NOS 755.280 Other specified reduction defect of upper limb 755.285 Transverse reduction defect of upper limb, NOS

755.290 Unspecified reduction defect of upper limb

Excludes shortening of lower limb (use 755.680) and hypoplasia of lower limb (use 755.685)

Includes: congenital amputation of upper limb, NOS

```
755.300 Absence of lower limb
                   femur (total or partial), tibia, fibula, and
         Absent:
         Includes: amelia of lower limb, NOS
                   infants with rudimentary or nubbin toes
                   attached to stump of femur or pelvic girdle
755.310 Absence of thigh and lower leg
        Absent:
                  femur (total or partial), tibia and fibula
                   (total or partial)
         Present: foot (total or partial)
         Includes: phocomelia of lower limb, NOS;
                   intercalary reduction defect of lower limb, NOS
755.320 Absence of lower leg only or femur only
         Absent: tibia and fibula
         Present: femur, foot (total or partial)
                   or
         Absent:
                  femur
         Present: tibia, fibula, and foot
755.330 Absence of lower leg and foot
         Absent: tibia and fibula (total or partial), foot
         Includes: infants with rudimentary or nubbin toes attached
                   to stump of leg or knee
755.340 Absence of foot or toes
                 foot or toes (total or partial) not in
         Absent:
                   conjunction with ray or long bone reduction
         Includes: rudimentary or nubbin toes;
                   absent individual phalanges;
                   absent or missing toes, NOS
         Excludes: isolated absent or hypoplastic great toe
                   (use 755.365)
         Split-foot malformation
755.350
                   central toes (third with or without second,
         Absent:
                   fourth) and metatarsals (total or partial)
         Includes: monodactyly;
                   lobster claw foot
         Excludes: isolated absent central toes without metatarsal
                   defects (use 755.340)
         Note: preaxial lower limb reductions can occur with
               split-hand malformations of the upper limb and these
               lower limb defects should be coded 755.365
755.360
        Longitudinal reduction defect of lower limb, NOS
         Includes: absent long bone of leg with absent toes, NOS
755.365
        Preaxial longitudinal reduction defect of lower limb
                  tibia (total or partial) and/OR great toe with
         Absent:
                   or without second toe (total or partial)
         Includes: isolated absent or hypoplastic great toe;
                   tibial ray defect, NOS
755.366 Postaxial longitudinal reduction defect of lower limb
         Includes: isolated absent fibula (total or partial);
                   absent fifth with or without fourth toe (total
                   or partial) only if fibula or fifth ± fourth
                   metatarsal also totally or partially absent;
                   fibular ray defect, NOS
755.380 Other specified reduction defect of lower limb
755.385 Transverse reduction defect of lower limb, NOS
               Includes: congenital amputation of lower limb, NOS
755.390 Unspecified reduction defect of lower limb
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755.4 Reduction defects of unspecified limb

T If description of condition includes amniotic or constricting bands use additional code, 658.800 (note: 658.00 should only be used with another reportable defect)

755.400 Absence of limb, NOS Includes: amelia, NOS

755.410 Phocomelia, NOS

Includes: intercalary reduction defect, NOS

755.420 Transverse reduction defect, NOS

Includes: congenital amputation of unspecified limb

755.430 Longitudinal reduction defect, NOS

Includes: preaxial or postaxial reduction defect, NOS

755.440 Absent digits, not specified whether fingers or toes

755.480 Other specified reduction defect of unspecified limb

755.490 Unspecified reduction defect of unspecified limb

755.5 Other anomalies of upper limb, including shoulder girdle

Includes: complex anomalies involving all or part of upper limb

755.500 Anomalies of fingers

Includes: camptodactyly clinodactyly

macrodactylia brachydactyly triphalangeal thumb incurving fingers

Excludes: acrocephalosyndactyly (see 756.050)

Apert's syndrome (see 756.055)

755.510 Anomalies of hand

Excludes: simian crease (use 757.200)

755.520 Anomalies of wrist 755.525 Accessory carpal bones

755.526 Madelung's deformity

755.530 Anomalies of forearm, NOS

755.535 Radioulnar dysostosis

755.536 Radioulnar synostosis

755.540 Anomalies of elbow and upper arm

755.550 Anomalies of shoulder

755.555 Cleidocranial dysostosis 755.556 Sprengel's deformity

755.560 Other anomalies of whole arm

755.580 Other specified anomalies of upper limb

Includes: hyperextensibility of upper limb

shortening of arm

755.585 Hypoplasia of upper limb

Includes: hypoplasia of fingers, hands, or arms

Excludes: aplasia or absent upper limb (see 755.2)

755.590 Unspecified anomalies of upper limb

755.6 Other anomalies of lower limb, including pelvic girdle

755.600 Anomalies of toes

Includes: overlapping toes

hammer toes

widely spaced first and second toes

755.605 Hallux valgus

755.606 Hallux varus

755.610 Anomalies of foot

Includes: plantar furrow

Excludes: lobster claw foot (use 755.350)

755.616 Rocker-bottom foot

755.620 Anomalies of ankle

astragaloscaphoid synostosis

755.630 Anomalies of lower leg

angulation of tibia, tibial torsion

(exclude if clubfoot present)

755.640 Anomalies of knee

hyperextended knee

755.645 Genu valgum

755.646 Genu varum

755.647 Absent patella or rudimentary patella

755.650 Anomalies of upper leg

anteversion of femur

755.660 Anomalies of hip

other abnormalities of hips

755.665 Hip dysplasia, NOS

755.666 Unilateral hip dysplasia

755.667 Bilateral hip dysplasia

755.670 Anomalies of pelvis

fusion of sacroiliac joint

755.680 Other specified anomalies of lower limb

hyperextended legs

shortening of legs

755.685 Hypoplasia of lower limb

Includes: hypoplasia of toes, feet, legs

Excludes: aplasia or absent lower limb (see 755.3)

755.690 Unspecified anomalies of legs

755.8 Other specified anomalies of unspecified limb

755.800 Arthrogryposis multiplex congenita

Includes: distal arthrogryposis syndrome

755.810 Larsen's syndrome

755.880 Other specified anomalies of unspecified limb

Includes: overlapping digits, NOS

hyperextended joints, NOS

Excludes: hyperextended knees (use 755.640)

755.9 Unspecified anomalies of unspecified limb

756 Other Congenital Musculoskeletal Anomalies

756.0 Anomalies of skull and face bones

Excludes: skull and face deformities in 754 Pierre Robin sequence (use 524.080) 756.000 Craniosynostosis, NOS craniostenosis, NOS closed-skull sutures, NOS 756.005 Sagittal craniosynostosis 756.006 Metopic craniosynostosis 756.010 Coronal craniosynostosis 756.020 Lambdoidal craniosynostosis 756.030 Other types of craniosynostosis Includes: basilar craniosynostosis 756.040 Craniofacial dysostosis Includes: Crouzon's disease 756.045 Mandibulofacial dysostosis Includes: Franceschetti syndrome Treacher-Collins syndrome 756.046 Other craniofacial syndromes Includes: oculomandibulofacial syndrome Hallermann-Streiff syndrome 756.050 Acrocephalosyndactyly, NOS 756.055 Acrocephalosyndactyly types I or II Apert syndrome 756.056 Acrocephalosyndactyly type III 756.057 Other specified acrocephalosyndactylies 756.060 Goldenhar syndrome oculoauriculovertebral dysplasia 756.065 Hemifacial microsomia 756.080 Other specified skull and face bone anomalies localized skull defects Includes: # flat occiput mid-facial hypoplasia # prominent occiput prominent maxilla hypotelorism Excludes: macrocephaly (use 742.400) small chin (see 524.0) Pierre Robin sequence (use 524.080) 756.085 Hypertelorism, telecanthus, wide set eyes 756.090 Unspecified skull and face bone anomalies Excludes: dentofacial anomalies (524.0) skull defects associated with brain anomalies such as: anencephalus (740.0) encephalocele (742.0) hydrocephalus (742.3) microcephalus (742.100)

756.1 Anomalies of spine

756.100 Spina bifida occulta 756.110 Klippel-Feil syndrome Wildervanck syndrome

756.120	Kyphosis
756.130	kyphoscoliosis Congenital spondylolisthesis
756.140	Anomalies of cervical vertebrae
756.145	Hemivertebrae (cervical)
756.146	Agenesis (cervical)
756.150	Anomalies of thoracic vertebrae
756.155	Hemivertebrae of thoracic vertebrae
756.156	Agenesis of thoracic vertebrae
756.160	Anomalies of lumbar vertebrae
756.165	Hemivertebrae of lumbar vertebrae
756.166	Agenesis of lumbar vertebrae
756.170	Sacrococcygeal anomalies
	Includes: agenesis of sacrum
756.179	Excludes: pilonidal sinus (see 685.100) Sacral mass, NOS
756.180	Other specified vertebral anomalies
756.185	Hemivertebrae, NOS
756.190	Unspecified anomalies of spine

756.2 Cervical rib

756.200 Cervical rib supernumerary rib in cervical region

756.3 Other anomalies of ribs and sternum

756.300	Absence of ribs				
756.310	Misshapen ribs				
756.320	Fused ribs				
756.330	Extra ribs				
756.340	Other anomalies of ribs				
756.350	Absence of sternum				
756.360	Misshapen sternum				
756.380	Other anomalies of sternum				
	Includes: double ossification center in the manubrium,				
	bifid sternum, short sternum				
756.390	Anomalies of thoracic cage, unspecified				
	Excludes: deformed chest (use 754.820)				

756.4 Chondrodystrophy

756.400	Asphyxiating thoracic dystrophy Jeune syndrome
	thoracic-pelvic-phalangeal dysplasia
	Excludes: homozygous achondroplasia
756.410	Chondrodysplasia
	Ollier syndrome, enchondromatosis
756.420	Chondrodysplasia with hemangioma
	Kast syndrome
	Maffucci syndrome
756.430	Achondroplastic dwarfism
756.440	Other specified chondrodystrophies
	Excludes: Conradi's (use 756.575)
756.445	Diastrophic dwarfism
756.446	Metatrophic dwarfism
756.447	Thanatophoric dwarfism

```
756.450 Metaphyseal dysostosis
756.460 Spondyloepiphyseal dysplasia
756.470 Exostosis
Excludes: Gardner syndrome (see 759.630)
756.480 Other specified chondrodystrophy
756.490 Unspecified chondrodystrophy
Excludes: lipochondrodystrophy (use 277.510)

756.5 Osteodystrophies

756.500 Osteogenesis imperfecta
756.505 Osteopsathyrosis
756.506 Fragilitas ossium
```

756.505 Osteopsathyrosis 756.506 Fragilitas ossium 756.510 Polyostotic fibrous dysplasia Albright-McCune-Sternberg syndrome 756.520 Chondroectodermal dysplasia 756.525 Ellis-van Creveld syndrome 756.530 Infantile cortical hyperostosis Caffey syndrome 756.540 Osteopetrosis Albers-Schonberg syndrome marble bones 756.550 Progressive diaphyseal dysplasia Engelmann syndrome Camurati-Engelmann disease 756.560 Osteopoikilosis 756.570 Multiple epiphyseal dysplasia 756.575 Conradi syndrome chondrodysplasia punctata Excludes: warfarin embryopathy

756.580 Other specified osteodystrophies 756.590 Unspecified osteodystrophies

756.6 Anomalies of diaphragm

756.600 Absence of diaphragm
756.610 Congenital diaphragmatic hernia
756.615 Diaphragmatic hernia (Bochdalek)
756.616 Diaphragmatic hernia (Morgagni)
756.617 Hemidiaphragm
756.620 Eventration of diaphragm
756.680 Other specified anomalies of diaphragm
756.690 Unspecified anomalies of diaphragm

756.7 Anomalies of abdominal wall

756.700 Exomphalos, omphalocele
756.710 Gastroschisis
Excludes: umbilical hernia (553.100)
756.720 Prune belly syndrome
756.790 Other and unspecified anomalies of abdominal wall
756.795 Epigastric hernia

756.8 Other specified anomalies of muscle, tendon, fascia and connective tissue

756.800 Poland syndrome or anomaly 756.810 Other absent or hypoplastic muscle

R = Rev. 6/07 N = Rev. 5/07 T = Rev. 6/04 * = code created by CDC # = on the MACDP Excl List Includes: absent pectoralis major
Excludes: prune belly syndrome (use 756.720)

756.820 Absent tendon

756.830 Nail-patella syndrome

756.840 Amyotrophia congenita

756.850 Ehlers-Danlos syndrome

756.860 Congenital torticollis
(see also 754.100, anomalies of sternocleidomastoid muscle)

756.880 Other specified anomalies of muscle, tendon, fascia and connective tissue
Includes: myopathy, congenital NOS

756.9 Unspecified anomalies of musculoskeletal system

756.900	Unspecified	anomalies	of	muscle
756.910	Unspecified	anomalies	of	tendon
756.920	Unspecified	anomalies	of	bone
756.930	Unspecified	anomalies	of	cartilage
756.940	Unspecified	anomalies	of	connective tissue
756.990	Unspecified	anomalies	of	musculoskeletal system

757 Congenital Anomalies of the Integument

757.000 Hereditary edema of legs Hereditary trophedema Milroy's disease

757.1 Ichthyosis congenita

757.100 Harlequin fetus
757.110 Collodion baby
757.115 Bullous type
757.120 Sjogren-Larsson syndrome
757.190 Other and unspecified
757.195 Ichthyosis vulgaris
757.196 X-linked ichthyosis
757.197 Ichthyosiform erythroderma

757.2 Dermatoglyphic anomalies

757.200 Abnormal palmar creases
Includes: simian creases, transverse palmar creases

757.3 Other specified anomalies of skin

Excludes:pigmented mole (216.900) hemangioma (see 228.0)

757.300 Specified syndromes, not elsewhere classified, involving skin anomalies

757.310 Skin tags

Includes: anal tags

Excludes: preauricular tag (see 744.110)

vaginal tags (see 752.480)

757.320 Urticaria pigmentosa

757.330 Epidermolysis bullosa

757.340 Ectodermal dysplasia

Excludes: Ellis-van Creveld syndrome (756.525)

757.345 X-linked type ectodermal dysplasia

757.346 Other specified ectodermal dysplasias

757.350 Incontinentia pigmenti

757.360 Xeroderma pigmentosum

757.370 Cutis laxa hyperelastica

757.380 Nevus, not elsewhere classifiable

Includes: port wine stain or nevus flammeus

T Excludes: hairy nevus (use 216.920)

Sturge-Weber syndrome (use 759.610)

757.385 Birthmark, NOS

757.386 Mongolian blue spot

757.390 Other specified anomalies of skin

skin cysts

hypoplastic dermal patterns

757.395 Absence of skin

757.4 Specified anomalies of hair

Excludes: kinky hair syndrome (use 759.870)

757.400 Congenital alopecia

Excludes: ectodermal dysplasia (use 757.340)

757.410 Beaded hair

Monilethrix

757.420 Twisted hair

Pili torti

757.430 Taenzer's hair

757.450 Persistent or excessive lanugo

Includes: hirsutism

757.480 Other specified anomalies of hair

757.5 Specified anomalies of nails

757.500 Congenital anonychia

Absent nails

757.510 Enlarged or hypertrophic nails

757.515 Onychauxis 757.516 Pachyonychia

757.520 Congenital koilonychia

757.530 Congenital leukonychia

757.540 Club nail

757.580 Other specified anomalies of nails

757.585 Hypoplastic (small) fingernails and/or toenails

757.6 Specified anomalies of breast

757.600 Absent breast with absent nipple

757.610 Hypoplastic breast with hypoplastic nipple

757.620 Accessory (ectopic) breast with nipple

757.630 Absent nipple

Т # 757.640 Small nipple (hypoplastic)

Always code if ≥36 weeks gestation

If <36 weeks gestation, code only if another reportable defect is present

757.650 Accessory (ectopic) nipple, supernumerary

757.680 Other specified anomalies of breast

Widely spaced nipples

Excludes: inverted nipples (never a defect)

757.8 Other specified anomalies of the integument

757.800 Includes: scalp defects

For specified anomalies of skin see 757.390

For specified anomalies of hair see 757.480

For specified anomalies of nails see 757.580

757.9 Unspecified anomalies of the integument

757.900 Unspecified anomalies of skin

757.910 Unspecified anomalies of hair, NOS

757.920 Unspecified anomalies of nail, NOS

757.990 Unspecified anomalies of the integument, NOS

758 Chromosomal Anomalies

758.0 Down syndrome

Clinical Down syndrome karyotype identified as:

- Т 758.000 Down syndrome, karyotype trisomy 21, cytogenetics result in record
- Т 758.008 Down syndrome suspected, cytogenetics pending
 - 758.010 Down syndrome, karyotype trisomy G, NOS

 - 758.020 Translocation trisomy duplication of a 21 758.030 Translocation trisomy duplication of a G, NOS
 - 758.040 Mosaic Down syndrome
- Т 758.090 Down syndrome, NOS (i.e. chart states a diagnosis of Trisomy 21 or Downs syndrome, but no cytogenetics result in record)
- Т 758.098 Down syndrome suspected, cytogenetics never done

758.1 Patau syndrome

Clinical Patau syndrome karyotype identified as:

- Т 758.100 Patau syndrome, karyotype trisomy 13, cytogenetics result in record
 - 758.108 Patau syndrome suspected, cytogenetics pending
 - 758.110 Patau syndrome, karyotype trisomy D, NOS
 - 758.120 Translocation trisomy duplication of a 13
 - 758.130 Translocation trisomy duplication of a D, NOS
- Т 758.190 Patau syndrome, NOS (i.e. chart states a diagnosis of Trisomy 13 or Patau syndrome, but no cytogenetics result in record)
- Т 758.198 Patau syndrome suspected, cytogenetics pending

758.2 Edwards syndrome

Clinical Edwards syndrome karyotype identified as:

- Т 758.200 Edwards syndrome, karyotype trisomy 18, cytogenetics result in record
- Т 758.208 Edwards syndrome suspected, cytogenetics pending
 - 758.210 Edwards syndrome, karyotype trisomy E, NOS

 - 758.220 Translocation trisomy duplication of an 18 758.230 Translocation trisomy duplication of an E, NOS
- 758.290 Edwards syndrome, NOS (i.e. chart states a diagnosis of Т Trisomy 18 or Edwards syndrome, but no cytogenetics
 - result in record) 758.295 Edwards phenotype - normal karyotype
- Т 758.298 Edwards syndrome suspected, cytogenetics pending

758.3 Autosomal deletion syndromes

```
758.300 Antimongolism syndrome
         Clinical antimongolism syndrome:
           karyotype - partial or total deletion of:
                 21
                 G, NOS
           NOS
758.310 Cri du chat syndrome
         Clinical Cri du chat syndrome:
           karyotype - deletion of:
                 5
                 B, NOS
           NOS
758.320 Wolff-Hirschorn syndrome
         Clinical Wolff-Hirschorn syndrome:
           karyotype - deletion of:
                 4
                 B, NOS
           NOS
758.330 Deletion of long arm of 13
deletion of long arm of D, NOS 758.340 Deletion of long arm of E \,
         deletion of long arm of 17 or 18
758.350 Deletion of short arm of E
         deletion of short arm of 17 or 18
758.360 Monosomy G mosaicism
758.370 Deletion in band 11 of long arm of 22 (22g11 deletions)
         Note: Code added for use with births on or after 4/1/2001
758.380 Other loss of autosomal material
758.390 Unspecified autosomal deletion syndromes
```

758.4 Balanced autosomal translocation in normal individual

758.400 Balanced autosomal translocation in normal individual

758.5 Other conditions due to autosomal anomalies

```
758.500 Trisomy 8
758.510 Other trisomy C syndromes
        Trisomy: 6, 7, 9, 10, 11, 12, or C, NOS
758.520 Other total trisomy syndromes
        Trisomy 22
        Trisomy, NOS
758.530 Partial trisomy syndromes
758.540 Other translocations
                     balanced translocation in normal
        Excludes:
                      individual (use 758.400)
758.580 Other specified anomalies of autosomes, NOS
        Includes:
                      marker autosome
758.585 Polyploidy
758.586 Triploidy
758.590 Unspecified anomalies of autosomes
```

758.6 Gonadal Dysgenesis

Excludes:pure gonadal dysgenesis (752.720)
Noonan syndrome (759.800)

758.600 Turner's phenotype, karyotype 45, X [XO]

758.610 Turner's phenotype, variant karyotypes

karyotype characterized by:

isochromosome

mosaic, including XO

partial X deletion

ring chromosome

Excludes: Turner's phenotype, karyotype normal XX

(use 759.800, Noonan syndrome)

758.690 Turner syndrome, karyotype unspecified, NOS

Bonneville-Ullrich syndrome, NOS

758.7 Klinefelter syndrome

758.700 Klinefelter's phenotype, karyotype 47, XXY

758.710 Klinefelter's phenotype, other karyotype with additional

X chromosomes

XX

XXXY

XXYY

XXXXY

758.790 Klinefelter syndrome, NOS

758.8 Other conditions due to sex chromosome anomalies

758.800 Mosaic XO/XY, 45X/46XY

Excludes: with Turner's phenotype (758.610)

758.810 Mosaic XO/XX

Excludes: with Turner's phenotype (758.610)

758.820 Mosaic XY/XXY,46XY/47XXY

Excludes: Klinefelter's phenotype (758.710)

758.830 Mosaic including XXXXY, 49XXXXY

Excludes: with Klinefelter's phenotype (use 758.710)

758.840 XYY, male, 47XYY

mosaic XYY male

758.850 XXX female, 47XXX

758.860 Additional sex chromosomes, NOS

758.880 Other specified sex chromosome anomaly

Includes: fragile X

758.890 Unspecified sex chromosome anomaly

758.9 Conditions due to anomaly of unspecified chromosomes

758.900 Mosaicism, NOS

758.910 Additional chromosome(s), NOS

758.920 Deletion of chromosome(s), NOS

758.930 Duplication of chromosome(s), NOS

758.990 Unspecified anomaly of chromosome(s)

759 Other and Unspecified Congenital Anomalies

759.0 Anomalies of spleen

```
759.000 Absence of spleen
asplenia
759.005 Ivemark syndrome
759.010 Hypoplasia of spleen
# 759.020 Hyperplasia of spleen
splenomegaly
hepatosplenomegaly (also use code 751.620)
759.030 Misshapen spleen
759.040 Accessory spleen
759.050 Ectopic spleen
759.080 Other specified anomalies of spleen
759.090 Unspecified anomalies of spleen
```

759.1 Anomalies of adrenal gland

759.100	Absence of adrenal gland
759.110	Hypoplasia of adrenal gland
759.120	Accessory adrenal gland
759.130	Ectopic adrenal gland
759.180	Other specified anomaly of adrenal gland
	Excludes: congenital adrenal hyperplasia
	(use 255.200)
759.190	Unspecified anomalies of adrenal gland

759.2 Anomalies of other endocrine glands

759.200	Anomalies of pituitary gland
759.210	Anomalies of thyroid gland
759.220	Thyroglossal duct anomalies
	thyroglossal cyst
759.230	Anomalies of parathyroid gland
759.240	Anomalies of thymus
	thymic hypertrophy
	absent thymus
759.280	Other specified anomalies of endocrine gland
759.290	Unspecified anomaly of endocrine gland
	759.210 759.220 759.230 759.240 759.280

759.3 Situs inversus

759.300	Dextrocardia with complete situs inversus
759.310	Situs inversus with levocardia
759.320	Situs inversus thoracis
759.330	Situs inversus abdominis
759.340	Kartagener syndrome (triad)
759.390	Unspecified situs inversus
	Excludes: dextrocardia (746.800) not
	associated with complete situs inversus

759.4 Conjoined twins

759.400	Dicephalus
	two heads
759.410	Craniopagus

head-joined twins 759.420 Thoracopagus thorax-joined twins 759.430 Xiphopagus xiphoid- and pelvis-joined twins 759.440 Pygopagus buttock-joined twins

759.480 Other specified conjoined twins

759.490 Unspecified conjoined twins

759.5 Tuberous sclerosis

759.500 Tuberous sclerosis Bourneville's disease epiloia

759.6 Other hamartoses, not elsewhere classified

759.600 Peutz-Jeghers syndrome 759.610 Encephalocutaneous angiomatosis Kalischer's disease Sturge-Weber syndrome 759.620 Von Hippel-Lindau syndrome 759.630 Gardner syndrome 759.680 Other specified hamartomas 759.690 Unspecified hamartomas

759.7 Multiple congenital anomalies,

759.700 Multiple congenital anomalies, anomaly, multiple, NOS deformity, multiple, NOS

759.8 Other specified anomalies and syndromes

759.800 Cong malformation syndromes affecting facial appearance cyclops Noonan syndrome oral-facial-digital (OFD) syndrome, type I Orofaciodigital syndrome, type II (Mohr syndrome) Waardenburg syndrome whistling face syndrome 759.820 Cong malformation syndromes associated with short stature

Amsterdam dwarf (Cornelia de Lange syndrome) Cockayne syndrome Laurence-Moon-Biedl syndrome

Russell-Silver syndrome

Seckel syndrome

Smith-Lemli-Opitz syndrome

759.840 Cong malformation syndromes involving limbs Carpenter syndrome Holt-Oram syndrome Klippel-Trenaunay-Weber syndrome Rubinstein-Taybi syndrome sirenomelia

thrombocytopenia-absent radius (TAR) syndrome

759.860 Cong malformation syndromes with other skeletal changes

R = Rev. 6/07N = Rev. 5/07T = Rev. 6/04* = code created by CDC # = on the MACDP Excl List Marfan syndrome

Stickler syndrome

759.870 Cong malformation syndromes with metabolic disturbances

Alport syndrome

Beckwith (Wiedemann-Beckwith) syndrome

leprechaunism

Menkes syndrome (kinky hair syndrome)

Prader-Willi syndrome Zellweger syndrome

759.890 Other specified anomalies

Includes: hemihypertrophy

Meckel-Gruber syndrome

759.9 Congenital anomaly, unspecified

759.900 Anomalies of umbilicus

low-lying umbilicus

umbilical cord atrophy

759.910 Embryopathia, NEC 759.990 Congenital anomaly, NOS

```
List ordered alphabetically
       524.000 Abnormalities of jaw size
               micrognathia
               macrognathia T
      255.200 Adrenogenital syndrome
      270.200 Albinism
    # 277.620 Alpha-1 antitrypsin deficiency
    # 658.800 Amniotic bands (constricting bands, amniotic cyst)
    # 270.600 Arginosuccinic aciduria
    # 778.000 Ascites, congenital
216
      Benign neoplasm of skin
    T (NOTE: All neoplasms should be coded ONLY if another reportable code
        is present)
                Includes: blue nevus
                                         pigmented nevus
                          papilloma
                                          dermatofibroma
                          syringoadenoma hydrocystoma
                        * dermoid cyst
                                         syringoma
               Excludes: skin of female genital organs (use 221.000),
                          skin of male genital organs (use 222.000)
T
               Benign neoplasm of skin, ear and external auditory canal
    # 216.200
                Includes: auricle ear
                           external meatus
                           auricular canal
                           external canal
                           pinna
                             cartilage of ear
               Excludes:
Т
    # 216.100
               Benign neoplasm of skin, eyelid, including canthus
               Excludes:
                             cartilage of eyelid
Τ
    # 216.000
               Benign neoplasm of skin, lip
               Excludes:
                             vermillion border of lip
    # 216.700 Benign neoplasm of skin, lower limb, hip
Т
    # 216.300
               Benign neoplasm of skin, other and unspecified parts of face
               Includes:
                             cheek, external nose, external eyebrow
                             temple
Т
    # 216.800
              Benign neoplasm of skin, other specified sites of skin
               Excludes:
                             epibulbar dermoid cyst (use 743.810)
T
    # 216.400
               Benign neoplasm of skin, scalp and skin of neck
Т
    # 216.900
               Benign neoplasm of skin, site unspecified
               Benign neoplasm of skin, trunk, except scrotum
    # 216.500
                             axillary fold
                Includes:
                             perianal skin
                             skin of:
                                                 wall,
                                                        abdominal wall,
                                          chest
                                          buttock, anus, perineum, back, umbilicus,
                                          breast
                             anal canal
               Excludes:
                             anus, NOS
                             skin of scrotum
```

```
T
      216.600
               Benign neoplasm of skin, upper limb, shoulder
               Benign skin neoplasm of female genital organs
      221.000
      222.000 Benign skin neoplasm of male genital organs
      453.000 Budd-Chiari, occlusion of hepatic vein
      427.900 Cardiac arrhythmias, NEC. Never code premature atrial
               contractions, PACs.
     330.100 Cerebral lipidoses
                            Tay-Sachs disease, gangliosidosis
               Includes:
      363.200
               Chorioretinitis
      279.200
               Combined immunodeficiency syndrome
      771.280
               Congenital infection, other specified
                             human immunodeficiency virus (HIV)
               Excludes:
                             infection and acquired immunodeficiency
                             syndrome (AIDS)
    # 277.000 Cystic fibrosis
               No mention of meconium ileus
     277.010 Cystic fibrosis
               With mention of meconium ileus
      228.100 Cystic hygroma
               Lymphangioma, any site
      771.100 Cytomegalovirus (CMV) (in utero infections only)
      253.820 Diencephalic syndrome
      279.110
               DiGeorge syndrome
      277.400 Disorders of bilirubin excretion
      425.300 Endocardial fibroelastosis
      553.200
               Epigastric hernia
      767.600
               Erb's palsy
      368.000
               Esotropia
      378.000
               Exotropia
      351.000
               Facial palsy
      331.890 Familial degenerative CNS disease
      760.710 Fetal alcohol syndrome
      760.718 Fetal alcohol syndrome, probable
               Includes: "facies"
      760.750 Fetal hydantoin (Dilantin) syndrome
      282.200 Glucose-6-phosphate dehydrogenase (G-6-PD) deficiency
      271.000 Glycogen storage diseases
Τ
      216.920 Hairy nevus
228.0 Hemangioma
                Include if greater than 4-inches diameter, if multiple
               hemangiomas, or if cavernous hemangioma
      228.040
               Hemangioma, intra-abdominal (Always code regardless of size, type or
               number)
               Hemangioma, intracranial (Always code regardless of size, type or
      228.020
      228.090
               Hemangioma, of other sites (Always code regardless of size, type or
               number)
     228.000
               Hemangioma, of unspecified site. Always code if multiple hemangiomas
               of any size are present, if one or more cavernous hemangiomas of any
               size are present, or if a single hemangioma measuring \geq 4cm in
               diameter or desribed as large, huge, or of medical significance is
               present.
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R = Rev. 6/07 N = Rev. 5/07 T = Rev. 6/04 * = code created by CDC # = on the MACDP Excl List

Т

228.030 Hemangioma, retinal (Always code regardless of size, type or number)

```
# 228.010
               Hemangioma, skin & subcutaneous, NOS Always code if multiple
               hemangiomas of any size are present, if one or more cavernous
               hemangiomas of any size are present, or if a single hemangioma
               measuring ≥ 4cm in diameter or desribed as large, huge, or of medical
               significance is present.
    # 286.000 Hemophilia (all types)
      774.490 Hepatitis, neonatal, NOS
      774.480 Hepatitis, neonatal, other specified
     282.100 Hereditary elliptocytosis
    # 282.000 Hereditary spherocytosis
      771.220 Herpes simplex (in utero infections only)
               Includes: encephalitis
                      meningoencephalitis
      202.300 Histiocytosis, malignant
      277.510 Hurler syndrome
               Includes:
                            lipochondrodystrophy
     778.600 Hydrocele, congenital
     270.700 Hyperglycinemia
     251.200 Hypoglycemia, idiopathic
     252.100 Hypoparathyroidism, congenital
     275.330 Hypophosphatemic rickets
      253.280 Hypopituitarism, congenital
              Hypothyroidism, congenital (Exclude even if other
    # 243.990
               defects are present only if the record specifies
               hypothyroidism of prematurity. Other types of
               hypothyroidism or hypothyroidism NOS should continue
               to be on the routine exclusion list.)
      345.600 Infantile spasms, congenital
    # 550.000 Inquinal hernia or patent processus vaginalis never
        -550.900 code in infants if <36 weeks gestation regardless of
                 the presence of a reportable defect.
                 NOTE: for those ≥36 weeks:
                 Code in males only if another reportable defect is present;
                 Code in females, always code even if found in isolation
      208.000 Leukemia, congenital, NOS
214
      Lipoma
      214.300 Lipoma, intra-abdominal organs
               Lipoma, intrathoracic organs
      214.200
      214.810 Lipoma, lumbar or sacral lipoma
                       paraspinal lipoma
      214.100 Lipoma, other skin and subcutaneous tissue
      214.800 Lipoma, other specified sites
      214.000 Lipoma, skin and subcutaneous tissue of face
      214.400 Lipoma, spermatic cord
      214.900 Lipoma, unspecified site
    # 457.800 Lymphatics - other specified disorders of (including chylothorax)
      524.000 Macrognathia
     270.300 Maple syrup urine disease
     777.600 Meconium peritonitis
     777.100 Meconium plug syndrome
      524.000 Micrognathia
      352.600 Moebius syndrome
```

```
# 520.600 Natal teeth
239.200 Neck cyst
774.490 Neonatal hepatitis, NOS
```

```
774.480 Neonatal hepatitis, other specified
      159.800 Neoplasms of the abdomen, other specified
      191.000 Neoplasms of the CNS
               Includes:
                            medulloblastoma, gliomas
      171.800 Neoplasms of the connective tissue
               Includes: Ewing's sarcoma
                             fibrosarcoma
      155.000 Neoplasms of the liver
               Includes:
                            hepatoblastoma
               hemangio-epithelioma
      162.800 Neoplasms of the lung
      186.000 Neoplasms of the testes
      194.000 Neuroblastoma
      237.700 Neurofibromatosis
     379.500 Nystagmus
    # 270.100 Phenylketonuria (PKU)
    * 524.080 Pierre Robin sequence
    # 685.100 Pilonidal sinus (sacrodermal), sacral sinus, sacral dimple
    # 277.630 Pseudocholinesterase enzyme deficiency
    # 284.000 Red cell aplasia
      362.600 Retinal degeneration, peripheral
      362.700 Retinitis pigmentosa
      190.500 Retinoblastoma
      771.000 Rubella, congenital (in utero infections only)
    # 685.100 Sacral dimple
    # 216.910 Sebaceous cyst
# 282.600 Sickle cell anemia
Τ
    # 090.000 Syphilis, congenital (in utero infections only)
      238.030 Teratoma, abdomen
      238.010 Teratoma, head and face
      238.020 Teratoma, neck
      238.000 Teratoma, NOS
      238.080 Teratoma, other specified
      238.040 Teratoma, sacral, coccygeal
      257.800 Testicular feminization syndrome
      771.090 TORCH infection, unspecified (in utero infections only)
    # 608.200 Torsion of the testes or spermatic cord
      771.210 Toxoplasmosis (in utero infections only)
    # 553.100 Umbilical hernia
    # 286.400 von Willebrand disease
      335.000 Werdnig-Hoffman disease
      189.000 Wilms tumor (nephroblastoma)
      426.705 Wolfe-Parkinson-White syndrome, congenital
```

List ordered by 6-digit code number

```
# 090.000
                     Syphilis, congenital (in utero infections only)
            155.000 Neoplasms of the liver
                     Includes:
                                   hepatoblastoma
                                   hmangio-epithelioma
            159.800 Neoplasms of the abdomen
            162.800 Neoplasms of the lung
            171.800 Neoplasms of connective tissue
                     Includes:
                                   Ewing's sarcoma
                                   fibrosarcoma
            186.000 Neoplasms of the testes
            189.000 Wilms tumor (nephroblastoma)
            190.500 Retinoblastoma
            191.000 Neoplasms of the CNS
                     Includes:
                                   gliomas
                                   mdulloblastoma
            194.000 Neuroblastoma
            202.300 Histiocytosis, malignant
            208.000 Leukemia, congenital, NOS
      214
                     Lipoma
            214.000 Lipoma, skin and subcutaneous tissue of face
             214.100 Lipoma, other skin and subcutaneous tissue
            214.200 Lipoma, intrathoracic organs
            214.300 Lipoma, intra-abdominal organs
            214.400 Lipoma, spermatic cord
            214.800 Lipoma, other specified sites
            214.810 Lipoma, lumbar or sacral lipoma
                     paraspinal lipoma
            214.900 Lipoma, unspecified site
T
      216
                     Benign neoplasm of skin
                      (NOTE: All benign neoplasms should be coded ONLY if
                       another reportable code is present)
                                   blue nevus pigmented nevus
                      Includes:
                                   papilloma
                                               dermatofibroma
                                   syringoadenoma
                                   *dermoid cyst
                                   hydrocystoma
                                   syringoma
                     Excludes:
                                   skin of female genital organs (use 221.000),
                                   skin of male genital organs (use 222.000)
          # 216.000
                    Skin of lip
                     Excludes:
                                   vermillion border of lip
          # 216.100 Eyelid, including canthus
                                   cartilage of eyelid
                     Excludes:
          # 216.200 Ear and external auditory canal
                     Includes:
                                   auricle ear
                                   external meatus
                                   auricular canal
                                   external canal
                                   pinna
                                   cartilage of ear
                     Excludes:
```

R = Rev. 6/07 N = Rev. 5/07 T = Rev. 6/04 * = code created by CDC

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216.400
                     Scalp and skin of neck
            216.500 Skin of trunk, except scrotum
                           Includes:
                                         axillary fold
                                         perianal skin
                              skin of:
                                         chest wall
                                         abdominal wall
                                         groin
                                         buttock
                                         anus
                                         perineum
                                         back
                                         umbilicus
                                         breast
                      Excludes:
                                   anal canal
                                   anus, NOS
                                   skin of scrotum
            216.600
                    Skin of upper limb, shoulder
            216.700
                     Skin of lower limb, hip
            216.800 Other specified sites of skin
                      Excludes:
                                   epibulbar dermoid cyst (use 743.810)
            216.900 Site unspecified
            216.910 Sebaceous cyst
             216.920 Hairy nevus
            221.000 Benign skin neoplasm of female genital organs
            222.000 Benign skin neoplasm of male genital organs
T
      228.0
                      Hemangioma
                      Include if greater than 4-inches diameter, if multiple
                      hemangiomas, or if cavernous hemangioma
            228.000
                      Hemangioma, of unspecified site
                      Always code if multiple hemangiomas of any size are present, if one
                      or more cavernous hemangiomas of any size are present, or if a single
                      hemangioma measuring ≥ 4cm in diameter or desribed as large, huge, or
                      of medical significance is present.
                     Hemangioma, skin & subcutaneous, NOS
          # 228.010
                      Always code if multiple hemangiomas of any size are present, if one
                      or more cavernous hemangiomas of any size are present, or if a single
                      hemangioma measuring ≥ 4cm in diameter or desribed as large, huge, or
                      of medical significance is present.
             228.020 Hemangioma, intracranial (Always code regardless of size, type or
                      number)
             228.030
                     Hemangioma, retinal (Always code regardless of size, type or number)
             228.040 Hemangioma, intra-abdominal (Always code regardless of size, type or
                      number)
             228.090 Hemangioma, of other sites (Always code regardless of size, type or
                     number)
             228.100 Cystic hygroma
                      Lymphangioma, any site
             237.700 Neurofibromatosis
             238.000 Teratoma, NOS
             238.010 Teratoma, head and face
             238.020 Teratoma, neck
             238.030 Teratoma, abdomen
             238.040 Teratoma, sacral, coccygeal
```

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# 243.990 Hypothyroidism, congenital
           (Exclude even if other defects are present only if the record
            specifies hypothyroidism of prematurity <36 weeks. Include other
            types of hypothyroidism and hypothyroidism NOS only when another
            reportable defect is present regardless of gestational age)
 251.200 Hypoglycemia, idiopathic
# 252.100 Hypoparathyroidism, congenital
  253.280 Hypopituitarism, congenital
  253.820 Diencephalic syndrome
  255.200 Adrenogenital syndrome (adrenal hyperplasia)
  257.800 Testicular feminization syndrome
 270.100 Phenylketonuria (PKU)
 270.200 Albinism
# 270.300 Maple syrup urine disease
# 270.600 Arginosuccinic aciduria
# 270.700 Hyperglycinemia
# 271.000 Glycogen storage diseases
# 275.330 Hypophosphatemic rickets
 277.000 Cystic fibrosis with no mention of meconium ileus
 277.010 Cystic fibrosis with mention of meconium ileus
  277.400 Disorders of bilirubin excretion
  277.510 Hurler syndrome
          Includes: lipochondrodystrophy
 277.620 Alpha-1 antitrypsin deficiency
 277.630 Pseudocholinesterase enzyme deficiency
  279.110 DiGeorge syndrome
  279.200 Combined immunodeficiency syndrome
 282.000 Hereditary spherocytosis
 282.100 Hereditary elliptocytosis
 282.200 Glucose-6-phosphate dehydrogenase (G-6-PD) deficiency
 282.600 Sickle cell anemia
 284.000 Red cell aplasia
 286.000 Hemophilia (all types)
 286.400 von Willebrand disease
 330.100 Cerebral lipidoses
          Includes:
                       Tay-Sachs disease
                       gangliosidosis
  331.890 Familial degenerative CNS disease
  335.000 Werdnig-Hoffman disease
  345.600 Infantile spasms, congenital
 351.000 Facial palsy
  352.600 Moebius syndrome
  362.600 Retinal degeneration, peripheral
  362.700 Retinitis pigmentosa
  363.200 Chorioretinitis
 368.000 Esotropia
 378.000 Exotropia
 379.500 Nystagmus
  425.300 Endocardial fibroelastosis
  426.705
          Congenital Wolfe-Parkinson-White syndrome
  427.900 Cardiac arrhythmias, NEC. Never code premature atrial
           contractions, PACs.
  453.000 Budd-Chiari, occlusion of hepatic vein
# 457.800 Other specified disorders of lymphatics (including chylothorax)
```

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# 520.600 Natal teeth
      524.000 Abnormalities of jaw size
               micrognathia
               macrognathia
    * 524.080 Pierre Robin sequence
    # 550.000- Inguinal hernia or patent processus vaginalis never
       550.900 code in infants if <36 weeks gestation regardless of the
               presence of a reportable defect.
               NOTE: for those ≥36 weeks:
                Code in males only if another reportable defect is present;
                in females, always code even if found in isolation
     553.100 Umbilical hernia
      553.200 Epigastric hernia
    # 608.200 Torsion of testes or spermatic cord
   # 658.800 Amniotic bands (constricting bands, amniotic cyst)
    \# 685.100 Pilonidal sinus (sacrodermal), sacral sinus, sacral dimple
      760.710 Fetal alcohol syndrome
      760.718 Probable fetal alcohol syndrome
               Includes:
                            "facies"
      760.750 Fetal hydantoin (Dilantin) syndrome
    # 767.600 Erb's palsy
771
               Congenital infections (in utero infections only)
               Excludes: congenital syphilis (use 090.000)
      771.000 Rubella, congenital
      771.090 TORCH infection, unspecified
      771.100 Cytomegalovirus (CMV)
      771.210 Toxoplasmosis
      771.220 Herpes simplex
               Includes:
                           encephalitis
                            meningoencephalitis
      771.280 Congenital infection, other specified
                           human immunodeficiency virus (HIV) infection and
               Excludes:
                            acquired immunodeficiency syndrome (AIDS)
      774.480 Hepatitis, neonatal, other specified
      774.490 Hepatitis, neonatal, NOS
     777.100 Meconium plug syndrome
     777.600 Meconium peritonitis
     778.000 Ascites, congenital
    # 778.600 Hydrocele, congenital
```

HHS:PHS:CDC:NCBDDD:DBDDD:06/16/04 Doc. 6digit88, Version 06/04

EXCLUSION LIST for the MACDP Nonreportable birth defects

Conditions Never to be Reported

The following newborn and infant conditions include those descriptions considered to be excludable or nonreportable conditions in the MACDP. This includes certain biochemical disorders not considered part of the present MACDP case definition.

Alphabetical list of conditions that are never considered to be defects.

Description

Anal fissure Atrial contractions, premature Breast hypertrophy Bronchopulmonary dysplasia (Wilson-Mikity syndrome) Cephalohematoma Chalasia (gastroesophageal reflux) CNS hemorrhage Conjunctivitis Diastasis recti Epulis Gastroesophageal reflux Gum cysts - Includes epulis, ranula, mucocele Hydrocephalus secondary to intraventricular hemorrhage (IVH) or CNS bleed Hip click, with no follow-up or therapy T Heart murmur Hyaline membrane disease Intestinal obstruction - requires chart review to determine if T cause of obstruction is a reportable defect. If so, code only T Intussusception - requires chart review to determine if cause of intussusception is a reportable defect. If so, code only the cause. Inverted nipples Laryngotracheomalacia or tracheomalacia Meconium stained skin or nails Mucocele Neonatal acne Overriding (overlapping) sutures Petechiae Phimosis Pneumothorax Premature atrial contractions Protruding tongue Ranula Redundant foreskin Retractile testes Tracheomalacia T Volvulus - requires chart review to determine if cause of volvulus is a reportable defect. If so, code only the cause. Wilson-Mikity syndrome R = Rev. 6/07

N = Rev. 5/07T = Rev. 6/04* = code created by CDC # = on the MACDP Excl List

EXCLUSION LIST for the MACDP Nonreportable birth defects

Conditions Which may be Included Under Certain Conditions

The following newborn and infant conditions include those descriptions considered to be excludable or nonreportable conditions in the MACDP, <u>but which may be included</u> under certain circumstances.

The following rules apply to coding these conditions:

- A. If a condition or defect listed appears in a chart, singly or in any combination with other defects listed only on the Exclusion List, <u>do not</u> fill out the case record form.
- B. If one of these conditions listed accompanies a reportable birth defect (from the 6-digit code manual and not on the exclusion list), then use the listed 6-digit code and record <u>all</u> defects (including those from this list) from the hospital record onto the case abstraction form.

<u>Alphabetical</u> list of conditions requiring \underline{no} record abstraction to be performed \underline{unless} associated with a reportable defect. The addition or revision dates of the changes in the list of conditions requiring no record abstraction are shown.

Revised/
Changed
Date

Changed				
Date			Code	Description
	T		744.100 757.650 270.200 277.620 658.800	Accessory auricle Accessory nipple (supernumerary nipple, or skin tag) Albinism Alpha 1-antitrypsin deficiency Amniotic bands (constricting bands, amniotic cyst)
10/1/92	T		757.310 746.400	Anal tags Aortic valve insufficiency or regurgitation, congenital - Code cases designated as 'mild', minimal', 'trivial', or 'physiologic' only if another reportable defect is present. Code all other degrees of insufficiency or regurgitation, including those where the degree is not specified, regardless of whether another reportable defect is present. Argininosuccinic aciduria
	T		778.000 744.220	Ascites or anasarca, congenital. Includes: hydrops fetalis Bat ear
	T	#	216.200	Benign neoplasm of skin, ear and external auditory canal Includes: auricle ear external meatus auricular canal external canal pinna Excludes: cartilage of ear
	T	#	216.100	Benign neoplasm of skin, eyelid, including canthus Excludes: cartilage of eyelid
	T	#	216.000	Benign neoplasm of skin, lip Excludes: vermillion border of lip

R = Rev. 6/07 N = Rev. 5/07 T = Rev. 6/04

^{* =} code created by CDC # = on the MACDP Excl List

T	#	216.700	Benign neoplasm of skin, lower limb, hip
T	#	216.300	Benign neoplasm of skin, other and unspecified parts of face
			Includes: cheek, external nose, external eyebrow, temple

EXCLUSION LIST for the MACDP Nonreportable birth defects

Alphabetical - Conditions Which may be Included Under Certain Conditions

Revised/ Changed	,			
<u>Date</u>			Code	Description
	T	#	216.800	Benign neoplasm of skin, other specified sites of skin Excludes: epibulbar dermoid cyst (use 743.810)
	T	#	216.400	Benign neoplasm of skin, scalp and skin of neck
	T	#	216.900	Benign neoplasm of skin, site unspecified
		#	216.500	Benign neoplasm of skin, trunk, except scrotum Includes: axillary fold perianal skin skin of: chest wall, abdominal wall, groin, buttock, anus, perineum, back, umbilicus, breast
				Excludes: anal canal, anus, NOS skin of scrotum
		#	216.600	Benign neoplasm of skin, upper limb, shoulder
			221.000	Benign skin neoplasm of female genital organs
			222.000 754.020	Benign skin neoplasm of male genital organs Bent nose, deviation of nasal septum
			744.820	Big lips
			757.385	Birth mark, NOS
			743.450	Blue sclera - if <36 weeks gestation, code only if
				another reportable defect is present. Always code if ≥36 weeks gestation.
			743.800	Brushfield spots
			757.390	Cafe au lait spots
			746.860	Cardiomegaly, congenital NOS
			744.230	Cauliflower ear
			330.100	Cerebral lipidoses (e.g., Tay-Sachs, gangliosidoses, etc.)
			756.200	Cervical rib
1 /1 /00			755.500	Clinodactyly (incurving of fifth finger)
1/1/93			752.520	Cryptorchidism (see undescended testicle)
			277.010	Cystic fibrosis, with mention of meconium ileus
			277.000 744.280	Cystic fibrosis, with no mention of meconium ileus Darwin's tubercle
1/1/96	T		754.030	Dolichocephaly - if <36 weeks gestation, code only if another reportable defect is present. Always code if
				≥36 weeks gestation.
1/1/93			743.800	Downward eye slant (antimongoloid)
			744.110	Ear tags, preauricular
			744.120	Ear tags, other
			744.230	Elfin ear, absent or decreased ear cartilage - if <36 weeks gestation, code only if another reportable defect is present.
			743.800	Epicanthal folds
			767.600	Erb's palsy
			368.000	Esotropia
			378.000	Exotropia

R = Rev. 6/07 N = Rev. 5/07T = Rev. 6/04

^{* =} code created by CDC # = on the MACDP Excl List

Revised 5/07 (EXCL1088) Replaces 6/93 Exclusion List

	351.000	Facial palsy
	757.380	Flammeus nevus or port wine stain
	748.180	Flat bridge of nose
	754.040	Fontanelle (large or small)
T	743.630	Fused eyelids - never code if <25 weeks gestation unless another reportable defect is present

EXCLUSION LIST for the MACDP Nonreportable birth defects

Alphabetical - Conditions Which may be Included Under Certain Conditions

Revised/ Changed

Date	Code	Description
	752.440 282.200 271.000 746.990	Fusion of vulva Glucose-6-phosphate dehydrogenase (G-6-PD) deficiency Glycogen storage disease Heart murmur - if chart review does not confirm a heart defect within 6 months, do not code as a defect even if other codable defects are present
3/4/91	286.000 751.620 282.100 282.000 750.240	Hemophilia Hepatomegaly Hereditary elliptocytosis Hereditary spherocytosis High arched palate
3/4/91	778.600 752.480 270.700 251.200 252.100 275.330	Hydrocele, congenital Hymenal tags Hyperglycinemia Hypoglycemia, idiopathic Hypoparathyroidism, congenital Hypophosphatemic rickets
1/1/96 T	752.440	Hypoplastic labia majora - if <36 weeks gestation, code only if another reportable defect is present. Always code if ≥36 weeks gestation.
3/4/91 T	748.510 752.810	Hypoplasia of lung; pulmonary hypoplasia - exclude only if an isolated defect in infants <36 weeks gestation Hypoplastic scrotum - exclude if secondary to undescended testes
T	243.990 752.430	Hypothyroidism, congenital (Exclude hypothyroidism of prematurity in infants <36 weeks gestation even if other reportable defects are present. Include other types of hypothyroidism and hypothyroidism NOS when another reportable defect is present regardless of gestational age) Imperforate hymen
Т	755.500 550.000- 550.900 550.901 550.902	Incurving fingers (clinodactyly) Inguinal hernia or patent processus vaginalis. Never code in infants <36 weeks gestation regardless of the presence of a reportable defect. For infants ≥36 weeks:
D. D. (07	757.450 754.040	<pre>In males, code only if another reportable defect is present; In females, always code even if found in isolation Lanugo, excessive or persistent Large fontanelle</pre>

R = Rev. 6/07 N = Rev. 5/07 T = Rev. 6/04

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Revised 5/07 (EXCL1088) Replaces 6/93 Exclusion List

	755.500	Long fingers and toes
	744.230	Lop ear
	744.245	Low set ears
	744.820	Macrocheilia (big lips)
	270.300	Maple syrup urine disease
	751.010	Meckel's diverticulum
	777.600	Meconium peritonitis
	777.100	Meconium plug
9/10/90	754.520	Metatarsus varus or adductus
		EXCLUSION LIST for the MACDP
		Management and the last of the state of the

Nonreportable birth defects

Alphabetical - Conditions Which may be Included Under Certain Conditions

Revi	sed/
Chan	ged

Date		Code	<u>Description</u>
		744.830	Microcheilia (small lips)
10/1/92	T	746.600	Mitral valve insufficiency or regurgitation, congenital -
		757.386 743.650	Code cases designated as 'mild', minimal', 'trivial', or 'physiologic' only if another reportable defect is present. Code all other degrees of insufficiency or regurgitation, including those where the degree is not specified, regardless of whether another reportable defect is present. Mongolian spots Nasal lacrimal duct obstruction
		520.600	Natal teeth
		745.500 379.500	Nonclosure of foramen ovale, NOS (see PFO) Nystagmus
9/10/90		756.080	Occiput, flat or prominent
3/5/90		457.800	Other specified disorder of lymphatics, including chylothorax
10/14/00	т	755.600	Overlapping toes
10/14/92	T	747.000	Patent ductus arteriosus (PDA) 1) Always code if ≥36 weeks of gestation and defect last noted at ≥6 weeks of age. 2) If ≥36 weeks gestation and defect last noted <6 weeks of age, code only if the PDA was treated)e.g. by ligation or indomethicin) or if another reportable defect is present. 3) Never code if <36 weeks gestation or if treated with prostaglanding regardless of gestational age.
10/14/92 T		745.500	Nonclosure of foramen ovale, NOS Patent foramen ovale (PFO) 1) Always code if ≥36 weeks of gestation and defect last noted at ≥6 weeks of age. 2) If ≥36 weeks gestation and defect last noted <6 weeks of age, code only if another reportable defect is present. 3) Never code if <36 weeks gestation regardless of presence of other defects.
	T	753.700	Patent urachus
8/1/93		744.820 747.325	Patulous lips (wide lips) Peripheral pulmonic stenosis (PPS) murmur - do collect
D. D.	0/07		-

R = Rev. 6/07 N = Rev. 5/07T = Rev. 6/04

^{* =} code created by CDC # = on the MACDP Excl List

	if PPS documented by echocardiogram
270.100	Phenylketonuria (PKU)
685.100	Pilonidal or sacral dimple
744.230	Pixie-like ear
744.230	Pointed ear
755.006	Polydactyly in blacks (postaxial, type B), includes
	only skin tags on hands or feet. All other types of
	postaxial polydactyly (i.e. extra finger with bone,
	nail, etc.) should always be coded.
744.246	Posteriorly rotated ears
744.410	Preauricular sinus, cyst or pit
744.110	Preauricular tags
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EXCLUSION LIST for the MACDP Nonreportable birth defects

Alphabetical - Conditions Which may be Included Under Certain Conditions

Revised/			
Changed			
Date		Code	Description
	T	747.680 752.450	Primary pulmonary artery hypertension Prominent clitoris
		277.630	Pseudocholinesterase enzyme deficiency
10/1/92	T	746.020	Pulmonary valve insufficiency or regurgitation, congenital Code cases designated as 'mild', minimal', 'trivial', or 'physiologic' only if another reportable defect is present. Code all other degrees of insufficiency or regurgitation, including those where the degree is not specified, regardless of whether another reportable defect is present.
		750.500	Pylorospasm (intermittent pyloric stenosis)
		751.580	Rectal fissures
		284.000	Red cell aplasia
		744.500	Redundant neck skin folds Rocker-bottom feet
		755.616 685.100	Sacral dimple
1 /1 /06	Т		
1/1/96		754.060	Scaphocephaly, no mention of craniosynostosis Always code if ≥ 36 weeks gestation.
	#		If <36 weeks gestation, code only if another reportable defect is present.
1/1/96	T	216.910 744.900 282.600 757.200 757.200 747.500	Sebaceous cysts Short neck Sickle cell anemia Sidney line Simian crease (transverse palmar crease) Single umbilical artery
		757.390 754.040 744.830	Skin cysts Small fontanelle
1/1/96	T	757.640	Small lips Small nipple (hypoplastic) Always code if ≥36 weeks gestation.
	#		If <36 weeks gestation, code only if another reportable defect is present
10/1/92 7/13/92	T	759.020 090.000 759.240	Splenomegaly Syphilis, congenital Thymic hypertrophy

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Revised 5/07 (EXCL1088) Replaces 6/93 Exclusion List

		755.630 750.000 608.200 608.200	Tibial torsion Tongue-tie Torsion of spermatic cord Torsion of testes
10/1/92	Т	746.105 759.900 553.100	Tricuspid valve insufficiency or regurgitation, congenital - Code cases designated as 'mild', minimal', 'trivial', or 'physiologic' only if another reportable defect is present. Code all other degrees of insufficiency or regurgitation, including those where the degree is not specified, regardless of whether another reportable defect is present. Umbilical cord atrophy Umbilical hernias (completely covered by skin)

EXCLUSION LIST for the MACDP Nonreportable birth defects

Alphabetical - Conditions Which may be Included Under Certain Conditions

Revised/ Changed Date		_Code	<u>Description</u>
1/1/93	T	752.500-	Undescended testicle (cryptorchidism)
	Т	748.180 743.800 752.460 752.480 286.400	1) If < 36 weeks gestation, code only if there is a medical/surgical intervention for this problem; 2) If ≥36 weeks gestation and defect last noted at <1 year of age, code only if there was a medical/surgical intervention for this problem or if another reportable defect is present 3) Always code if ≥36 weeks gestation and defect first noted at ≥1 of age. Upturned nose Upward eye slant (mongoloid) Vaginal cysts Vaginal tags von Willebrand's disease
3/14/91	Т	755.130 744.500 748.180 755.600 757.680	Webbed toes Code webbing of the second and third toes only if another reportable defect is present. Always code webbing of other toes regardless of whether another reportable defect is present Webbing of neck Wide nasal bridge Widely spaced first and second toes Widely spaced nipples

EXCLUSION LIST for the MACDP

 $\underline{\underline{\textbf{Numerical}}}$ list of conditions requiring \underline{no} record abstraction $\underline{\underline{\textbf{unless}}}$ associated with a reportable defect. The addition or revision dates of the changes in the list of conditions requiring no record abstraction are shown. .

Revised/ Changed

Date Date		Code	Description
7/13/92	216	090.000	Syphilis congenital Benign neoplasm of skin (NOTE: All benign neoplasms should be coded ONLY if another reportable code is present) Includes: blue nevus pigmented nevus papilloma dermatofibroma syringoadenoma *dermoid cyst hydrocystoma syringoma
		Exclude	
	#	216.000	Skin of lip
	#	216.100	Excludes: vermillion border of lip Eyelid, including canthus Excludes: cartilage of eyelid
	#	216.200	Ear and external auditory canal Includes: auricle ear
	#	216.300	Excludes: cartilage of ear Skin of other and unspecified parts of face
	II	210.300	Includes: cheek, external nose, external eyebrow, temple
	#	216.400	Scalp and skin of neck
	#	216.500	Skin of trunk, except scrotum Includes: axillary fold
	# # #	216.600 216.700 216.800	Excludes: anal canal anus, NOS skin of scrotum Skin of upper limb, shoulder Skin of lower limb, hip Other specified sites of skin Excludes: epibulbar dermoid cyst (use 743.810)

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#	216.900	Site unspecified
#	216.910	Sebaceous cyst

EXCLUSION LIST for the MACDP

 $\underline{\textbf{Numerical}}$ list of conditions requiring \underline{no} record abstraction \underline{unless} associated with a reportable defect. The addition or revision dates of the changes in the list of conditions requiring no record abstraction are shown. .

Revised/ Changed

Date		Code	Description
		221.000 222.000	Benign skin neoplasm of female genital organs Benign skin neoplasm of male genital organs
	T	243.990	Hypothyroidism, congenital (Exclude even if other defects are present only if the record specifies hypothyroidism of prematurity <36 weeks. Include other types of hypothyroidism and hypothyroidism NOS only when another reportable defect is present regardless of gestational age.
		251.200 252.100	Hypoglycemia, idiopathic Hypoparathyroidism, congenital
		270.100 270.200	Phenylketonuria (PKU) Albinism
		270.300	Maple syrup urine disease
		270.600	Argininosuccinic aciduria
		270.700	Hyperglycinemia
		271.000 275.330	Glycogen storage diseases
		277.000	Hypophosphatemic rickets Cystic fibrosis, with no mention of meconium ileus
		277.010	Cystic fibrosis, with mention of meconium ileus
		277.620	Alpha 1-antitrypsin deficiency
		277.630	Pseudocholinesterase enzyme deficiency
		282.000	Hereditary spherocytosis
		282.100	Hereditary elliptocytosis
		282.200	Glucose-6-phosphate dehydrogenase (G-6-PD) deficiency
		282.600	Sickle cell anemia
		284.000 286.000	Red cell aplasia Hemophilia
		286.400	von Willebrand's disease
		330.100	Cerebral lipidoses (e.g., Tay-Sachs, gangliosidoses, etc.)
		351.000	Facial palsy
		368.000	Esotropia
		378.000	Exotropia
		379.500	Nystagmus
3/5/90		457.800	Other specified disorder of lymphatics, including chylothorax
		520.600	Natal teeth
	T	550.000-	Inguinal hernia or patent processus vaginalis never
		550.900	code in infants if <36 weeks gestation regardless of the presence of a reportable defect. NOTE: for those ≥ 36 weeks:
			in males , code only if another reportable defect is present;
		FF2 100	in females , always code even if found in isolation
D	Rev. 6/07	553.100	Umbilical hernias (completely covered by skin)
r =	1167.0/07		

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	608.200 608.200	Torsion of spermatic cord Torsion of testes			
T	658.800 685.100	Amniotic bands (constricting bands, amniotic cyst) Pilonidal or sacral dimple			
		EXCLUSION LIST for the MACDP			
		Nonreportable birth defects			

Numerical - Conditions Which may be Included Under Certain Conditions

Revised/
Changed
Data

Changed		G = 4 =	Posseriation
<u>Date</u>		<u>Code</u>	Description
	T	743.450	Blue sclera - if <36 weeks gestation, code only if another reportable defect is present. Always code if ≥36 weeks gestation.
	T	743.630	Fused eyelids - never code if <25 weeks gestation unless another reportable defect is present
		743.650	Nasal lacrimal duct obstruction
		743.800	Brushfield spots
		743.800	Downward eye slant (antimongoloid)
		743.800	Epicanthal folds
		743.800	Upward eye slant (mongoloid)
		744.100	Accessory auricle
		744.110	Ear tags, preauricular
		744.120	Ear tags, other
		744.220	Bat ear
		744.230	Cauliflower ear
		744.230	Elfin ear, absent or decreased ear cartilage
			If <36 weeks gestation, code only if another
			reportable defect is present.
		744.230	Lop ear
		744.230	Pixie-like ear
		744.230	Pointed ear
		744.245	Low set ears
		744.246	Posteriorly rotated ears
		744.280	Darwin's tubercle
		744.410	Preauricular sinus, cyst or pit
		744.500	Redundant neck skin folds
		744.500	Webbing of neck
		744.820	Macrocheilia (big lips)
		744.820	Patulous lips (wide lips)
		744.830 744.900	Microcheilia (small lips) Short neck
		745.500	Nonclosure of foramen ovale, NOS (see PFO)
10/11/00	Т		
10/14/92	1	745.500	Patent foramen ovale (PFO) 1)Always code if ≥36 weeks of gestation and defect last
			noted at ≥ 6 weeks of age.
			2) If ≥36 weeks gestation and defect last noted <6 weeks
			of age, code only if another reportable defect is
			present.
			3) Never code if <36 weeks gestation regardless of
			presence of other defects.
10/1/92	T	746.020	Pulmonary valve insufficiency or regurgitation,
			congenital -
			Code cases designated as 'mild', minimal', 'trivial',
			or 'physiologic' only if another reportable defect is
	0/07		

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present. Code all other degrees of insufficiency or regurgitation, including those where the degree is not specified, regardless of whether another reportable defect is present.

EXCLUSION LIST for the MACDP Nonreportable birth defects

Numerical - Conditions Which may be Included Under Certain Conditions

Revised/ Changed			
Date		Code	<u>Description</u>
10/1/92	T	746.105	Tricuspid valve insufficiency or regurgitation, congenital - Code cases designated as 'mild', minimal', 'trivial', or 'physiologic' only if another reportable defect is present. Code all other degrees of insufficiency or regurgitation, including those where the degree is not specified, regardless of whether another reportable defect is present.
10/1/92	T	746.400	Aortic valve insufficiency or regurgitation, congenital - Code cases designated as 'mild', minimal', 'trivial', or 'physiologic' only if another reportable defect is present. Code all other degrees of insufficiency or regurgitation, including those where the degree is not specified, regardless of whether another reportable defect is present.
10/1/92	T	746.860 746.860 746.990	Mitral valve insufficiency or regurgitation, congenital - Code cases designated as 'mild', minimal', 'trivial', or 'physiologic' only if another reportable defect is present. Code all other degrees of insufficiency or regurgitation, including those where the degree is not specified, regardless of whether another reportable defect is present. Cardiomegaly, congenital NOS Heart murmur - if chart review does not confirm a
		740.990	heart defect within 6 months, do not code as a defect even if other codable defects are present
10/14/92	T	747.000	Patent ductus arteriosus (PDA) 1) Always code if ≥36 weeks of gestation and defect last noted at ≥6 weeks of age. 2) If ≥36 weeks gestation and defect last noted <6 weeks of age, code only if the PDA was treated)e.g. by ligation or indomethicin) or if another reportable defect is present. 3) Never code if <36 weeks gestation or if treated with prostaglandins regardless of gestational age.
8/1/93		747.325 747.500 747.680	Peripheral pulmonic stenosis (PPS) murmur - do collect if PPS documented by echocardiogram Single umbilical artery Primary pulmonary artery hypertension

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Revised 5/07 (EXCL1088) Replaces 6/93 Exclusion List

		778.000 748.180 748.180 748.180	Ascites or anasarca. Includes: hydrops fetalis Flat bridge of nose Upturned nose Wide nasal bridge
3/4/91	T	748.510	Hypoplasia of lung; pulmonary hypoplasia - exclude if isolated defect in infants <36 weeks gestation.
		750.000	Tonque-tie
3/4/91		750.240	High arched palate
		750.500	Pylorospasm (intermittent pyloric stenosis)
		751.010	Meckel's diverticulum
			EXCLUSION LIST for the MACDP

Nonreportable birth defects

Numerical - Conditions Which may be Included Under Certain Conditions

Revised/ Changed

Changed		Date	Code	Description
		751.580 751.620 752.430 752.440	Rectal fissur Hepatomegaly Imperforate h Fusion of vul	ymen
1/1/96	T	752.440	code only if	abia majora - if <36 weeks gestation, another reportable defect is present. f ≥36 weeks gestation.
3/4/91		752.450 752.460 752.480 752.480	Prominent cli Vaginal cysts Vaginal tags Hymenal tags	
1/1/93	T	752.500-	Undescended to	esticle (cryptorchidism)
	Т	752.520	medical/surgi 2) If ≥36 week year of age, intervention defect is pre	if \geq 36 weeks gestation and defect first
1/1/93		752.520 752.810 753.700 754.020	Cryptorchidis Hypoplastic s undescended to Patent urachu	m (see undescended testicle) crotum - exclude if secondary to estes
1/1/96	T	754.030	code only if	y - if <36 weeks gestation, another reportable defect is present. f ≥36 weeks gestation.
1/1/93 1/1/96		754.040 754.060	Fontanelle (1 Scaphocephaly If <36 weeks reportable de	arge or small) , no mention of craniosynostosis gestation, code only if another fect is present. f ≥36 weeks gestation.
1/1/93		754.520 755.006	Metatarsus va Polydactyly i only skin tag postaxial pol	rus or adductus n blacks (postaxial, type B), includes s on hands or feet. All other types of ydactyly (i.e. extra finger with bone, hould always be coded.

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Revised 5/07 (EXCL1088) Replaces 6/93 Exclusion List

3/14/91	T	755.130 755.500 755.500	Webbed toes Code webbing of the second and third toes only if another reportable defect is present. Always code webbing of other toes regardless of whether another reportable defect is present Clinodactyly (incurving of fifth finger) Long fingers and toes
		755.600	Overlapping toes
		755.600	Widely spaced first and second toes
		755.616	Rocker-bottom feet
		755.630	Tibial torsion
		756.080	Occiput, flat or prominent
		756.200	Cervical rib

EXCLUSION LIST for the MACDP Nonreportable birth defects

Numerical - Conditions Which may be Included Under Certain Conditions

	Revised/ Changed			
	Date		Code	Description
			757,200	Sidney line
			757.200	Simian crease (transverse palmar crease)
			757.310	Anal tags
			757.380	Flammeus nevus or port wine stain
			757.385	Birth mark, NOS
			757.386	Mongolian spots
			757.390	Cafe au lait spots
			757.390	Skin cysts
			757.450	Lanugo, excessive or persistent
	1/1/96	T	757.640	Small nipple (hypoplastic)
	_, _, _,	_		If <36 weeks gestation, code only if another
				reportable defect is present.
				Always code if ≥36 weeks gestation.
	9/10/90		757.650	Accessory nipple (supernumerary nipple, or skin
tag)	-, -,			
5,			757.680	Widely spaced nipples
			759.020	Splenomegaly
			759.240	Thymic hypertrophy
			759.900	Umbilical cord atrophy
			767.600	Erb's palsy
			777.100	Meconium plug
			777.600	Meconium peritonitis
			778.000	Ascites or anasarca, congenital
			778.600	Hydrocele, congenital

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T = Rev. 6/04

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MACDP Decision Tree for Determining Whether to Include Patent Ductus Arteriosus (PDA)

```
Is the child on
prostaglandins?
              ----> Yes ---->
                                           Never code
     ω
   No
     ω
What was the gestational
age of the child at birth? -----> < 36 wks ---> Never code
     ω
   \geq 36 wks
     ω
How old was the
child when defect
was last noted? ----> > 6 wks ---> Always code
    ω
   < 6 wks
     \omega
Has the PDA been
treated? (e.g., by
ligation or
                    ----> Yes ----> Always code
indomethicin)
     \omega
   No
Include only if another reportable heart defect is present.
```

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MACDP Decision Tree for Determining Whether to Include Patent Foramen Ovale (PFO)

R = Rev. 6/07 N = Rev. 5/07 T = Rev. 6/04 * = code created by CDC # = on the MACDP Excl List

MACDP Decision Tree for Determining Whether to Include Peripheral Pulmonary Stenosis (PPS)

May 22, 1996

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6.1 Introduction

The National Birth Defects Prevention Network (NBDPN) is committed to improving the quality, accuracy, completeness, comparability, and timeliness of birth defects surveillance data. Information on the prevalence of birth defects reported by surveillance systems can vary considerably due to differences in case definition, method of case ascertainment, and the types of data sources used.

This chapter describes two major approaches to birth defects surveillance: active case ascertainment and passive case ascertainment. The *active case ascertainment* approach is the intensive level of case identification that involves staff finding cases at strategic data sources. Ascertainment is usually very complete, and each diagnosis in the database is confirmed. In the *passive case ascertainment* approach the surveillance program receives case reports of birth defects from data sources. The completeness of ascertainment is highly dependent on the number and types of data sources used by the surveillance program and on the consistency of case reporting from the data sources. Since case reports usually are not confirmed by staff in a passive case ascertainment program, it is particularly important for these programs to implement quality assurance procedures aggressively.

Although the two surveillance approaches are operationally different, it is possible to achieve comparable levels of data quality. Programs should evaluate their surveillance approaches regularly for accuracy, completeness, and timeliness and should be creative in identifying strategic means of quality improvement.

In this chapter we first introduce some relevant terminology (Section 6.2). We then discuss general surveillance development (Section 6.3) and introduce approaches to case identification (Section 6.4). In Sections 6.5 and 6.6 we present in some detail information on the two main approaches to case identification (active and passive case ascertainment, respectively). The remaining sections cover additional topics in case ascertainment, including data sources (Section 6.7), sources of information that may be available at a given data source (Section 6.8), and issues relating to infant risk factors and case identification (Section 6.9). References cited in this chapter may be found in Section 6.10.

Appendices to this chapter provide additional detail on the following important data sources for birth defects surveillance: vital records (Appendix 6.1), hospital data sets (Appendix 6.2), hospital and patient services logs (Appendix 6.3), and genetic services (Appendix 6.4).

Chapter 6 6-1 Case Ascertainment Methods

6.2 Terminology

Surveillance (public health)

The ongoing, systematic collection, analysis, and interpretation of health data essential to the planning, implementation and evaluation of public health practice, closely integrated with the timely dissemination of these data to those who need to know (Centers for Disease Control and Prevention, 1988).

Monitoring

The performance and analysis of routine measurements using statistical methods aimed at detecting changes in the environment or health status of populations (Last, 1995).

Registry

A system of ongoing registrations, such that cases of a disease or other health-relevant condition are defined in a population and can be related to a population base. Birth and death registration systems are examples. Some disease registries, like the cancer registry, closely resemble public health surveillance systems and have epidemiologic value (Last, 1995).

Case ascertainment or identification

The process of identifying – from existing sources and using defined case definitions – embryos, fetuses, neonates, infants, and children who have a birth defect.

Active case ascertainment

A surveillance approach to case identification that is based on surveillance staff being engaged intensively in all activities related to finding and confirming potential birth defects cases. Surveillance staff seek out data sources and conduct systematic investigations of pertinent sources of information to identify potential cases of birth defects. Data collection sites include hospital medical records, diagnostic indices, unit logs, pathology departments, and specialty sites.

Passive case ascertainment

A surveillance approach to case identification whereby birth defects programs receive case reports from data sources. Passive case ascertainment programs receive case reports from one or many different data sources and may accommodate multiple reporting formats including hard copy, electronic, and web-based reports, as well as administrative data sets. There may be variability in the completeness and accuracy of case ascertainment in programs that use this type of case ascertainment.

Population-based surveillance

Surveillance that identifies a population under study, usually defined by geopolitical boundaries, and establishes the denominator from which cases come. A data source that is population based covers an entire population within a defined area. Some examples of population-based data sources are: vital records (birth, death, and fetal death), statewide newborn genetic screening programs, and statewide newborn hearing screening programs.

Data source

Any facility, site, or entity that has cases or potential cases of birth defects or other pertinent medical information. This includes a hospital, clinic, physician's office, laboratory, prenatal diagnosis center, as well as administrative databases.

Reportable disease

A disease, laboratory result, or health condition of public health significance that requires notification of its occurrence to a public health agency. Authorizing legislation or regulations usually define which conditions are reportable, which data sources are required to report, timelines for reporting, and what demographic information is required, at a minimum, in a case report.

Reporting source

A data source that is required, by law, to report or allow access to cases of birth defects and other pertinent medical conditions to the birth defects program.

Administrative data set

A data set or database that is created to fulfill operational or managerial objectives. Many are developed as information management systems with multiple functions. Examples include hospital discharge data, Medicaid data, vital records master index, clinical management information systems, health care billing and insurance claims systems.

Unit

A component, section, or department within a data source that serves a specific function or performs a specific activity. Examples include health information management department, labor and delivery unit, neonatal intensive care unit, and pathology department

Data collection

The process of gathering information, which includes receiving, retrieving, accessing, abstracting, and extracting information from information sources.

Medical records review (information review)

The process of reading, identifying, interpreting, and translating documentation per specific program objectives. Medical records review precedes abstracting.

Abstracting

The process of recording information, identified when reviewing documentation in a medical record or other information source, and entering the information into data fields in a specified format. Information may be recorded on hard copy forms or through formatted data entry computer screens.

Disease coding

The process of assigning a standardized disease code (e.g., ICD-9-CM or 6-digit CDC code) to medical information.

Case abstract or case record

The documentation file(s) containing complete information about the birth defects case.

6.3 General Surveillance Development

Birth defects surveillance systems should be developed to facilitate the essential activities of data collection, data analysis, data evaluation, and information dissemination consistent with a program's established goals and objectives. The general guidelines below can be applied to developing a new system or improving an existing system. We are indebted to Mausner and Bahn (1974) and Teutsch and Churchill (2000) for much of the information in this section.

In the following sections we discuss planning and documenting the system (Section 6.3.1), identifying data sources (Section 6.3.2), obtaining knowledge about individual data sources (Section 6.3.3), implementing data quality procedures (Section 6.4.3), and evaluating surveillance method and analytical capability (Section 6.3.5).

6.3.1 Plan and Document

A birth defects surveillance program must be organized and have supporting documentation before beginning operations. The program can begin to process case reports once the logistics of case identification and data collection are established with data sources. Therefore, it is important to engage surveillance staff, data sources, stakeholders, advisors, and others affected by program operations early in the planning process.

The program should:

- ➤ Understand the legal authority and restrictions that shape surveillance operations, including processes for changing or amending legislation (see Chapter 2 on Legislation).
- Develop a mission statement and define the surveillance program's goals and objectives. Determine what outcome measurements are desired by the program. For example, the program may want to describe the distribution of birth defects in their population, calculate rates and perform statistical analyses, and identify children who require services. Ideally, the development of goals, objectives, and outcome measurements will be done in collaboration with stakeholders and with internal and external advisory groups.
- ➤ **Define** the parameters of case definition for the surveillance program, including residency, pregnancy outcomes, eligible diagnoses, and age range. Define the minimum criteria for an eligible case report (see Chapter 3 on Case Definition).
- ➤ **Define** the method of case identification that will be used. Usually, a program will develop an infrastructure to support functions of active or passive case ascertainment. It is essential to document procedures, protocols, decision items, and methods of data collection (the program's surveillance approach). Records review and data collection procedures should be defined precisely.
- ➤ **Determine** the data variables needed to fulfill program goals and objectives. Define the minimum information that must be collected and address other information that would be beneficial to the surveillance program (see Chapter 4 on Data Variables).
- **Document** protocol and procedures regarding the privacy of the individual and the confidentiality of health information.

Chapter 6 6-4 Case Ascertainment Methods

- ➤ **Design** forms for reporting, data collection, and abstracting that are adaptable to computer technology. This could include web-based reporting and forms that provide for easy data entry or scanning and that support abstracting medical records in the field (see Chapter 9 on Data Management and Security).
- ➤ **Develop** a database that has record linkage capability and that also functions as an information management system. The database should be flexible, adaptable, and able to accept electronic transfer of data files, web-based case reports, and case record abstracts from multiple sources. The database should support identification of all sources of information through which a diagnosis is identified or reported. It is also useful to be able to track and monitor medical records requests and perform other information management functions (see Chapter 9 on Data Management and Security).

6.3.2 Identify Data Sources

A key component in surveillance is identifying data sources for case ascertainment. A program needs to understand and evaluate the traits, characteristics, and operating procedures of all data sources. This is particularly important if there are potential sources of bias or underreporting associated with the way cases may be identified at a source.

The program should:

- ➤ **Identify** all potential data sources able to provide information that will help to fulfill the program's mission (e.g., hospitals, genetics and specialty clinics, cytogenetics laboratories, administrative data sets, vital records).
- ➤ Determine which data sources are included in any legislation mandating reporting and any additional sources for voluntary reporting. Consideration should be given to recommending legislative changes if program objectives change or are expanded, or if important data sources are omitted from mandatory reporting. For example, when adding prenatal diagnosis surveillance to program operations, it may be necessary to amend legislative language to include new data sources or facilities

6.3.3 Obtain Knowledge about Individual Data Sources

For each reporting data source the program should:

- **Know** the data source's mission or goals.
- ➤ **Identify** professional or legal mandates governing operations of the data source that may affect access to, or quality of, data from that source.
- **Describe** the population served by the data source.
- ➤ Chart the flow of information that is relevant to the surveillance program through the unit and/or data source. This is a good way to determine how the information is collected originally and whether or not the information is forwarded to a central repository (e.g., centralized computer file, medical records department, administrative database).
- Maintain an up-to-date directory of names and contact information for relevant people at the data source (e.g., medical records personnel).

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- ➤ Utilize multiple data sources. Surveillance programs should use multiple data sources, both for case identification and data collection. It is important for the surveillance program to realize that one source rarely fills all of a surveillance system's needs for case record accuracy, completeness, and timeliness.
- ➤ **Develop** record linkage procedures to facilitate matching all reports to the correct case record. This is especially important when programs utilize multiple data sources (see Chapter 9 on Data Management and Security).

6.3.4 Implement Data Quality Procedures

Surveillance programs should evaluate data for completeness, accuracy, timeliness, and comparability to other birth defects programs. At a minimum, programs should develop quality assurance procedures (manual and/or computerized) to identify potential issues in data quality. This includes accuracy, completeness, and timeliness. Additionally, programs should maintain documentation on data collection, data abstraction, and medical records review procedures. This will reduce the risk of multiple interpretations that lead to an inconsistent application of procedures and interpretation of medical information. For further details, see Chapter 7 on Data Quality Management.

6.3.5 Evaluate Surveillance Method and Analytical Capability

Surveillance programs should evaluate the surveillance approach and determine whether the surveillance system is meeting program objectives. Additionally, outcome measurements should be evaluated. NBDPN recommends the guidelines offered in the document *Updated Guidelines for Evaluating Public Health Surveillance Systems* (Centers for Disease Control and Prevention, 2001).

Chapter 6 6-6 Case Ascertainment Methods

6.4 Birth Defects Surveillance – Approaches to Case Identification

Cases of birth defects are generally identified in one of two ways: through 'active case ascertainment' (i.e., staff conduct case finding) or through 'passive case ascertainment' (i.e., case reports are received by the program). While some surveillance systems use both kinds of ascertainment approaches for case identification, program activities are generally structured around one or the other approach.

Birth defects rates are directly related to the method of case identification and type of surveillance approach. Table 6.1 presents birth defects rates based on various surveillance approaches (Edmonds, 1997).

Table 6.1 Birth Defects Rates by Surveillance Approach

Data Source	% of Babies Reported with Birth Defects
Birth Certificates in 1996	1.5
Newborn Hospital Discharge Data (Florida)	4.3-7.1
Mandatory Hospital Reporting (New York)	3.4
Linked Data Sources (North Carolina)	4.7
Active Hospital Surveillance (Atlanta 1992-1996)	2.6
Physical Exam of Infants (Collaborative Perinatal	8.3
Project)	

Although a physical examination of each infant provides the most complete assessment of birth defects among births, and therefore the highest prevalence, few programs can sustain this type of intensive case ascertainment. At the other extreme, the prevalence at birth of defects is clearly underreported when only birth certificates are used in case ascertainment. The NBDPN promotes case ascertainment approaches that provide a more complete description of birth defects prevalence in the US.

Whereas the previous section on general surveillance development (Section 6.3) provides a foundation for surveillance systems, the following two sections (Sections 6.5 and 6.6) discuss unique issues that arise in using either active or passive case ascertainment approaches in the identification of birth defect cases. We are indebted to Lynberg and Edmonds (1992) for much of the information in Sections 6.5 and 6.6.

Chapter 6 6-7 Case Ascertainment Methods

6.5 Active Case Ascertainment

With active case ascertainment, cases of birth defects are identified at data sources by surveillance staff. The case-finding process includes identifying potential birth defects cases, reviewing and abstracting information from medical records, and conducting follow-up in order to complete abstracts or verify information. Programs take measures to ensure complete case ascertainment by using multiple data sources and multiple units within data sources. Case-finding activities may vary depending on the program's resources and objectives. A program's resources, as well as program goals and objectives, should be used to determine the intensity of case finding. Regardless of the case-finding methods used, active surveillance programs should provide detailed instructions on the case-finding process, document procedures for collecting information and completing case abstracts, nurture relationships between the program and its data sources, evaluate the quality and effectiveness of all steps in the case-finding process, and implement quality improvement methods.

In the sections below we discuss characteristics of active case ascertainment (Section 6.5.1), a recommended approach for active case ascertainment (Section 6.5.2), data quality issues in active case ascertainment (Section 6.5.3), evaluation (Section 6.5.4), and tips and hints for active case ascertainment (Section 6.5.5).

6.5.1 Characteristics of Active Case Ascertainment

- Surveillance staff identify birth defects cases by visiting data sources. Staff should follow a thorough and systematic set of investigative methods so that all potential birth defects cases are identified.
- > Surveillance staff are trained to find birth defects cases. Staff learn how to find (or *cull*) cases in hospitals, medical facilities, clinics, laboratories (e.g., cytogenetics laboratories, genetics clinics, prenatal diagnostic centers), and in medical records that relate to each potential case (e.g., prenatal, maternal delivery, newborn, infant, pediatric).
- > Staff are trained to gather information from information sources and medical records. This includes following abstracting procedures and documentation guidelines. Staff are trained in birth defects coding and learn how to conduct follow-up.
- Multiple information sources are used to obtain data. All potential data sources should be part of the case-finding investigative process, and some are essential (e.g., birth hospitals, unit logs in birth hospitals). Surveillance systems should evaluate the effectiveness of case finding at each data source.
- Case abstract forms are detailed and comprehensive and usually include a number of variables pertaining to the pregnancy, delivery, and outcome. Information on the mother and infant is often collected in detail, including medical and prenatal care history, complications of pregnancy or delivery, reproductive history, physical examinations, postnatal procedures, and birth defects diagnosis.
- Clinical reviewers, usually physicians, are trained to confirm, qualify, and evaluate the diagnostic information collected by the surveillance abstractors.
- Active case-finding surveillance should result in accurate and complete identification of birth defects cases. The data are of high quality due to extensive staff training. The data collected are comprehensive and result in a detailed case abstract.

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6.5.2 Approach to Active Case Ascertainment

Active surveillance is based on surveillance staff investigating data sources and finding potential birth defects cases. Although other activities may be part of the active surveillance approach, case finding is the primary task. There are various approaches to the case-finding process. Some programs have staff review all pertinent data sources and information reports, while others limit case finding to the most important information sources. Some use existing databases or lists of potential cases that are generated by the data source. Because case finding is labor intensive, most programs evaluate case-finding activities and determine ways to identify cases effectively and efficiently, yet still be relatively sure that case ascertainment is complete. It is necessary to take into account the legal or legislative issues that govern program activities.

Essential program activities for active case ascertainment include those listed below.

- ➤ Identify program objectives. It is important to develop or enhance the case-finding approach based on the purpose and objectives of the surveillance system. For example, if information is used to refer children to services, then the case-finding process should be designed to collect identifying and contact information early enough in the process to make the referral in a timely manner.
- ➤ **Develop** a flow chart of the case-finding process. Identify the data sources that are consistently used for case finding. At a minimum the program must conduct extensive case finding at birth and major pediatric hospitals. Within the data sources, the program should identify which units and departments will always be used. Important units and departments to consider are labor and delivery, nursery, surgery, and pathology (see list of data sources in Section 6.7). Some programs use the medical records department to generate a list of diagnoses (i.e., disease codes) from the disease index.
- ➤ **Define** the type of information to look for and collect during the case-finding process. Information gathered may be sketchy, incomplete, and general. This is especially true when gathering information from unit logs. The case-finding process may also include gathering information for the conditions of low birth weight, prematurity, and other conditions that may potentially lead to a case.
- ➤ **Define** the frequency of case-finding activities (i.e., visiting sources of information and completing abstracts). Frequency and consistency of case-finding activities affect the timeliness of the surveillance database. For example, if the program identifies a child who needs to be referred for services, it is usually important for the referral to be made in a timely manner. Timeliness can be measured by setting goals for the maximal length of time between birth and referral.
- Conduct case finding (culling). This is the systematic and ongoing process of identifying birth defects cases. Potential cases at the data source are found by surveillance staff through one or more procedures: (1) reviewing information at unit logs within a data source and creating a list of medical records to be pulled by the health information department within the data source; and/or (2) requesting a line listing of potential cases from the data source or unit, usually by identifying the cases by ICD codes (e.g., hospital index); or (3) reviewing the medical records for every delivery, termination, miscarriage, etc. occurring at the data source.

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- ➤ Conduct medical records reviews. Potential cases of birth defects identified by the case-finding process are further investigated through medical records reviews. Requests for medical records are provided to staff of medical records departments at hospitals (or other sites), who pull the charts and make them available to surveillance system staff. Surveillance staff, who determine if the child or fetus meets the eligibility criteria for inclusion as a case, review the medical records. Multiple medical records may be reviewed during this process. These may include: maternal medical records during prenatal care, hospital admits during the pregnancy, and the delivery record. Medical records for a child include the newborn delivery record and any medical (hospital) records generated after the birth.
- Abstract information. As medical records are reviewed, surveillance staff abstract (record) the required information and record it on the case abstract form. Trained surveillance staff follow program guidelines and procedures for completing the data elements on the case abstract, confirming a diagnosis, and conducting follow-up to find cases at data sources and within units at data sources. Although a surveillance program develops its own set of abstracting guidelines and procedures, these should be based on established guidelines when available. In some programs, the abstractor assigns the disease code. In others, assigning the disease code occurs separately from abstracting.
- Perform a clinical review. Some programs have an expert in medical diagnosis issues review the case abstract after it is complete. The abstract is evaluated for incomplete data variables (i.e., data fields), accuracy of the medical information, and accuracy of the disease code assigned. Some clinical reviews result in the further classification of the case with a summary diagnosis, as an isolated or syndromic case, or other classification.

6.5.3 Data Quality Issues in Active Case Ascertainment

Active case finding requires surveillance staff to review and collect information from medical records. Staff are involved directly in verifying and confirming medical information and determining whether further follow-up or investigation is needed. In these programs, the burden of maintaining the quality of the database rests with the surveillance staff. It is essential to understand the challenges to data quality that occur in active surveillance and to implement strategies to identify and to correct them (see also Chapter 7 on Data Quality Management).

- Field work (case finding, record review, abstracting) should be evaluated for accuracy, incomplete data variables, and consistency. Desired outcome benchmarks in each of these areas should be identified and improvements implemented and tracked.
- Data sources and individual units should be evaluated with respect to the staff resources expended and the results obtained. Since case finding is labor intensive, programs should streamline and improve operations whenever possible. The value of the output of each unit or department utilized should be evaluated against the staff resources used. The program should determine whether unnecessary medical records are being reviewed and identify which non-anomaly ICD codes are most effective in identifying potential cases.
- The surveillance database should be evaluated for timeliness. This includes measuring how current the database is in relation to calculating disease rates. Although programs may collect information on individual birth defects cases over many months or years, they should set benchmarks for finalizing an individual case record or meeting a level of productivity by specified times.

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6.5.4 Active Case Ascertainment Surveillance Evaluation

Evaluation of active ascertainment surveillance methods should occur at two levels. Both levels directly impact data quality and the program's ability to meet goals and objectives. One level targets case identification and data collection. Examples of areas that should be evaluated are:

- Completeness
- > Accuracy
- > Timeliness
- Measurability

Programs should develop outcome measurements that will improve data quality and are important to meet program needs and surveillance objectives. See Chapter 7 on Data Quality Management for a more detailed discussion of this topic.

The other level focuses on the surveillance system itself. For a comprehensive approach to evaluating surveillance systems refer to CDC's *Updated Guidelines for Evaluating Surveillance Systems* (Centers for Disease Control and Prevention, 2001).

6.5.5 Tips and Hints in Active Case Ascertainment

- Establish precise guidelines and criteria for data requests to data sources. The process of active case ascertainment includes requesting information from data sources. Specific criteria or data variable parameters should be provided (e.g., ad hoc reports generated from an existing database, extracted information from databases).
- Visit tertiary care (e.g., major pediatric site) hospitals first. These sites usually have the most complete diagnostic information on a birth defects case. Surveillance staff can follow the case back to the birth hospital for the remaining information. Often a delivery that occurs in a rural hospital is transferred to the tertiary care facility.
- Coordinate the schedule of site visits with the data sources to minimize inconvenience for them.
- Form relationships with staff in medical record departments (directors, coders, those coordinating release of information, record retrievers), birth registrar at the hospital, and hospital unit staff. Discuss the purposes of the surveillance program with them and describe the work that surveillance staff perform at their sites.
- Know key information technology or data processing staff at the data source. These individuals often can access and retrieve specific pieces of information collected at the data source or within a component unit.
- ➤ Use caution with ICD lists or an ICD disease index generated by medical records (or information technology) staff. Hospital medical records coders are required to adhere to a set of federal guidelines when assigning a disease code to the medical record. Surveillance staff use a set of abstracting guidelines developed by the program (or NBDPN). Measure the benefit of using a disease code from an index against the output gained and resources used. For example, evaluate the results of a medical records review after using disease codes from an index.
- ➤ Use laptops. Design computer screens to assist in the case-finding process. Direct data entry during information gathering is more efficient, and likely more accurate, than recording information on paper forms and then entering it into the database.

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➤ Be conscious of HIPAA, especially as this relates to the privacy and security rules that covered entities (i.e., health care facilities) are required to follow. Be knowledgeable about public health exemptions in HIPAA. Provide reassuring documentation to sites as appropriate (see Chapter 2 on Legislation).

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6.6 Passive Case Ascertainment

With passive case ascertainment, case reports are submitted by data sources to the surveillance program. The level of interaction between programs and reporting sources varies, as do the methods of reporting. Some programs create birth defects case reporting forms and instruct reporting sources on how to complete them. Other programs merge or extract pertinent information into the surveillance program's database from a data source's existing database. Many use a combination of reporting methods to develop as complete an identification of birth defects cases as possible within the resources available. Regardless of the methods used, operating a surveillance system that receives case reports from data sources requires the program to identify and use multiple data reporting sources, provide detailed instructions to case reporting sources, nurture the relationship between the program and the reporting data source, and evaluate the quality of the case reports received.

In the sections below we discuss characteristics of passive case ascertainment (Section 6.6.1), a recommended approach for passive case ascertainment (Section 6.6.2), data quality issues in passive case ascertainment (Section 6.6.3), evaluation (Section 6.6.4), and tips and hints for passive case ascertainment (Section 6.6.5).

6.6.1 Characteristics of Passive Case Ascertainment

- ➤ Birth defects cases are reported by data sources to the surveillance program.
- Medical information is received by the program as a case report and is generally accepted as reported (i.e., the program does not confirm every case report for accuracy or comprehensiveness of diagnostic information).
- The operational procedures used by each data source influence the accuracy, totality, definition, and timeliness of reported diagnoses. This, in turn, influences the quality of the data in the surveillance program's database.
- Information is usually reported from multiple reporting sources. Data sources often serve different purposes for a program. Many can be used as major sources of clinical information (e.g., hospital reports, hospital discharge index, cytogenetics laboratories). Some are used as a source of demographic and statistical information (e.g., vital records). Others are used primarily for tracking or follow-up (e.g., genetics clinics, pathology and autopsy reports, specialty treatment clinics, and developmental centers). See the list of data sources in Section 6.7.
- The database is developed to accommodate various reporting formats. Information may be submitted in many ways and formats, including web-based reporting, electronic transfer and digital format, computerized reports, and hard copy reporting forms. Medical information may be reported in text format or in ICD code.
- Record-matching procedures are used since data are collected from multiple sources and existing databases. Case report information is extracted from administrative databases (e.g., hospital discharge data set, Medicaid data, vital records) and from existing databases within a facility (e.g., laboratories, specialty clinics, prenatal diagnostic centers).

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6.6.2 Approach to Passive Case Ascertainment

Passive case ascertainment is based on case reports submitted to the program by data reporting sources. Reporting sources may include mandatory hospital reporting and physician reporting and/or administrative databases (e.g., hospital discharge data set, Medicaid data, vital records). Completeness in the surveillance system is improved by using multiple data sources, especially when data sources are selected to fill a gap in case identification (e.g., fetal death certificates, pathology and autopsy reports). Customized reporting forms may be used, or a program may elect to use other methods for receiving case reports. All legal or legislative issues that govern program operations must be taken into account.

Essential program activities for passive case ascertainment include those listed below.

- Establish the type and scope of passive case ascertainment that defines program operations, including whether surveillance includes fetal deaths. Some programs have limited disease reporting guidelines and a smaller set of data sources that are required to report. Some programs may have more liberal disease reporting guidelines but, due to limited resources, have to limit the scope of program operations. Generally, programs that use multiple data sources will have more complete case ascertainment than those that use only one or two data sources. If programs use the birth certificate as a data source for case reports, they should use another data source for case identification.
- ➤ Identify the case identification data sources. These include birth and major pediatric hospitals. If fetal death is an outcome that is ascertained, it is important to use the fetal death certificate, and possibly cytogenetics laboratories, as a source of case identification. See the list of data sources in Section 6.7.
- ➤ **Define** case reporting requirements precisely for each data source. This includes identifying the required or minimum data variables that should be reported. Some data sources will only report the required minimum data variables, while others, like an administrative database, may be able to furnish the program with additional pieces of information. Refer to Chapter 4 on Data Variables.
- ➤ **Develop** data reporting methods and procedures for each data source, including data format, timeliness, or reporting schedules. When possible, encourage electronic or web-based reporting. Data sources are usually more consistent in reporting when the burden of submitting the case report is minimized.
- **Develop** record linkage capability. It is important not only to accommodate multiple case reporting formats, but also to use the efficiencies of technology in processing case reports from administrative and existing databases and linking them to case records in the program's database.
- ➤ **Develop** procedures for abstracting information from medical records. This includes using the NBDPN Abstractor' Instructions (see Chapter 3 on Case Definition, Appendix 3.2), assigning disease codes, recording other pertinent information, and entering data into the database. Passive case ascertainment programs should review medical records as part of data quality evaluations. Additionally, medical records reviews are often conducted for other focused surveillance functions. For example, some programs that perform statistical monitoring regularly review medical records to confirm a diagnosis. Other times it is important for surveillance staff to review medical records to confirm a diagnosis during a community investigation or when investigating a suspected cluster.

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6.6.3 Data Quality Issues in Passive Case Ascertainment

In passive case ascertainment, reporting sources submit case reports to the surveillance system. The reports are accepted without prior confirmation or verification of the information. Therefore, evaluations for quality must be conducted, especially regarding key program outcomes such as completeness, accuracy, and timeliness. Evaluations are often done by reviewing medical records and comparing results between the review and the reported diagnosis. A result of the evaluation process should include quality assurance procedures to identify future problems and methods to track improvement (see also Chapter 7 on Data Quality Management).

- The quality of a reported diagnosis should be evaluated for accuracy and comprehensiveness. Errors and differences in reporting will occur, resulting in underreporting, overreporting, and inaccurate reporting. By "rating" the quality of a reported diagnosis, data sources can also be evaluated. Results can be used to adjust quality control and assurance procedures and direct strategic programmatic decisions.
- The surveillance database should be evaluated for timeliness. This includes measuring how 'current' the database is in relation to the program's ability to calculate disease rates. Track timeliness of reporting per data source and identify reporting time lags. For example, watch reporting trends to identify whether some calendar months or quarters are problematic for some data sources. Evaluate the surveillance program's data processing procedures for time lags.
- The disease coding classification system should be evaluated to identify weaknesses, limitations, and problematic codes. This is especially important for data sources that report cases in ICD code format, which can happen with a data source such as an administrative or existing database. Additionally, although federal coding guidelines are used to direct a hospital or clinic medical records coder in assigning a disease code, the interpretation of medical documentation in the chart is often the reason for a particular code assignment. A good way for a surveillance program to identify potential code problems is to understand some of the conditions that may surface during the newborn time period. For example, a problematic code could be 748.0, choanal atresia or stenosis, since some newborns do experience difficulty in breathing in the first few hours of life. Additionally, situations that might cause a misuse of codes are low birth weight and prematurity (see Chapter 3 on Case Definition, Appendix 3.3). To gain experience in understanding these issues, medical records should be reviewed and results evaluated
- The surveillance database should be evaluated for fluctuations in counts and rates of specific diagnoses. It is possible that an increase in a rate may be due to a change in procedure at a data source. Passive case ascertainment systems must understand that procedures and processes at the data source affect the quality of information in the surveillance database.
- The surveillance program should develop benchmarks for desired outcome measurements and develop strategies for how to improve the outcome results. For example, a critical data source that is consistently lagging in reporting might be the focus of a strategic plan to improve timeliness.

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6.6.4 Passive Case Ascertainment Surveillance Evaluation

Evaluation of passive case ascertainment surveillance methods should occur at two levels. Both levels directly impact data quality and the program's ability to meet goals and objectives.

One level targets data reporting sources, case identification, and data collection. Examples of areas that should be evaluated are:

- Completeness
- Accuracy
- > Timeliness
- Measurability

Programs should develop outcome measurements that will improve data quality and are important to meet program needs and surveillance objectives. See Chapter 7 on Data Quality Management for a more detailed discussion of this topic.

The other level focuses on the surveillance system itself. For a comprehensive approach to evaluating surveillance systems refer to CDC's *Updated Guidelines for Evaluating Surveillance Systems* (Centers for Disease Control and Prevention, 2001).

6.6.5 Tips and Hints in Passive Case Ascertainment

- ➤ Use record linkage to link to the vital record early in the data collection process. The vital records data source is excellent for establishing a unique case in the database and one that readily identifies the residency of the pregnancy outcome. Additionally, the birth and fetal death certificates fulfill many data collection variables for pregnancy outcome, maternal, and pregnancy information, as well as other statistical information (see Chapter 4 on Data Variables). See the detailed description of the vital records data source in Appendix 6.1.
- ➤ Identify high-quality data sources that report a confirmed diagnosis. A diagnosis from a high-quality source is an efficient way to improve the accuracy of the database. It also offsets the need to conduct a medical records review for quality evaluations for the specific diagnosis.
- Ensure cooperation and compliance of data sources as critical factors in passive case ascertainment. Ease the burden on data sources by encouraging electronic, computerized, and web-based reporting formats for submitting case reports. Offer technical assistance to sites. Many data sources already have the information the surveillance system needs in a database. It is usually easier to sustain consistent, timely, and compliant reporting using a computer program to extract information, rather than expecting staff at the data source to complete a case report.
- ➤ Be flexible when discussing reporting methods and reporting requirements with a data source. All data sources may not be able to provide all of the desired 'minimum' data fields easily. Evaluate the contribution, including efficiencies, the data source can make to the surveillance system and adjust reporting requirements accordingly. Identify which sources can usually be depended upon to report the majority of demographic information.
- ➤ Be knowledgeable about the information flow through respective hospitals and sites. Understand medical records content and documentation practices, including how the ICD code classification is used. Passive case ascertainment systems should be proactive in understanding where to go and who to contact to clarify issues when problems arise.

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- Consider conducting 'case finding' at a data source as an alternative to receiving the case report. Although 'case finding' is not part of the passive surveillance approach, this method should be considered for data sources that may not have an efficient or reliable method of reporting (e.g., outpatient specialty clinics), that may not be able to report in a thorough manner (e.g., autopsy/pathology), or that are not required to report (i.e., voluntary reporting).
- Communicate with data sources on how birth defects data are used. Identify the users of the data (the customers) and some of the products produced using surveillance information. Reporting sources like to be recognized for the contributions they make (i.e., reporting cases) and appreciate knowing that the data they provide are used and serve important and valuable purposes.
- ➤ Be active and creative in managing the quality of the database. It is possible to develop program strategies that not only promote the efficiencies of passive case ascertainment but also improve the important outcome measurements of accuracy, completeness, and timeliness.
- ➤ Be conscious of HIPAA, especially as this relates to the privacy and security rules that covered entities (i.e., health care facilities) are required to follow. Be knowledgeable about the public health exemptions in HIPAA. Provide reassuring documentation to sites as appropriate (see Chapter 2 on Legislation).

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6.7 Data and Case Identification Sources

Information on birth defects cases can be obtained from many sources, each of which has strengths and limitations. Rarely is one source able to provide all of the information necessary to complete a case record. Some, like birth and pediatric hospitals, are ideal for identifying a large number of cases. However, it is important not to overlook data sources like cytogenetics laboratories and specialty outpatient clinics, since they may identify cases previously unknown to a birth defects program. The challenge for birth defects surveillance programs is to evaluate and select data sources that meet the objectives of the program and that can be accessed using available resources. Most data sources can be useful for both active and passive case identification. Differences arise between the two case ascertainment approaches in how the information is gathered and collected. Some data sources are more conducive to active case ascertainment since the only way to access the information is to physically gather it. Some of the major data sources – including vital records, hospital discharge data, hospital unit logs, and genetics clinics – are described in further detail in Appendices 6.1, 6.2, 6.3, and 6.4.

Vital Records (see Appendix 6.1 for detailed description)

- ➤ Birth certificates
- > Fetal death certificates
- ► Elective termination reports
- Death certificates

Hospital Information (see Appendix 6.2 for detailed description)

- ➤ Hospital discharge data set
- ➤ Hospital disease index

Hospital Unit Logs, including (see Appendix 6.3 for detailed description):

- Labor and delivery
- Surgery
- Nursery
- ➤ Neonatal Intensive Care Unit (NICU)

Hospital Departments, including:

- Pathology
 Forensic (autopsy) pathology
 Surgical pathology
- Surgery
 Inpatient and outpatient/ambulatory

> Specialty and outpatient clinics

Obstetrics

Prenatal

Perinatology

Laboratory

Pediatric medicine

Prenatal and Obstetrics Centers

- Birthing centers
- Obstetrics services
- ➤ Planned Parenthood, and other women's care clinics
- ➤ Prenatal diagnosis and high-level ultrasound referral sites
- Prenatal genetics counseling services

Specialty Clinics

- ➤ Genetics (see Appendix 6.4 for detailed description)
- > Oral-facial, craniofacial
- ➤ Meningomyelocele
- Cardiology
- Pulmonary/respiratory
- ➤ Musculo-skeletal
- > Developmental and growth
- > Audiology and speech
- **Early** intervention
- ➤ Neuro-developmental
- Ophthalmology

Laboratories

- Cytogenetics
- > Prenatal diagnosis
- ➤ Metabolic

Physicians

- Pediatricians
- Obstetricians
- > Specialists

Health Care Professionals

- Audiologists
- > Developmental therapists

Administrative Databases

- > Statewide hospital discharge data set (see Appendix 6.2 for detailed description)
- ➤ Medicaid data
- ➤ HMO data sets

Other Sources of Information

- > University-based medical clinics
- ➤ Newborn hearing screening program
- > Newborn genetic screening program
- > Coroners and medical examiners
- ➤ Child fatality/mortality review programs
- Public health maternal and child health programs
 Public health clinics, including developmental clinics
- School records

6.8 Sources of Information in a Data Source

In this section we discuss the various sources of information that may be available at a given data source. In Section 6.8.1 we provide a general introduction to the medical record, followed by a more detailed discussion of the various types of documentation within a medical record in Section 6.8.2. Other sources of information discussed include maternal delivery medical records (Section 6.8.3); prenatal medical records (Section 6.8.4); cytogenetic laboratory reports (Section 6.8.5); and autopsy, pathology, and laboratory reports (Section 6.8.6).

6.8.1 Medical Records

By law, all health care facilities are required to maintain some form of medical record on every patient for every service encounter that occurs in the facility. A medical record provides documentation on the course of treatment and progress of the patient at the facility for each admission or service encounter. The medical record may also include information from other health care facilities that may be pertinent to the treatment at that facility. For additional information on the professional practices and standards for medical records and other issues related to health information management, please consult the American Health Information Management Association (http://www.ahima.org).

Medical records differ according to type of health care facility. Medical records maintained by a private health care provider, genetic counseling facility, hospital, or cytogenetics laboratory are likely to differ in the documentation included in the record and how the records are organized. The medical records that birth defects program staff are most likely to work with are those maintained by hospitals, particularly birth and tertiary care pediatric hospitals, and specialty clinics.

The documentation required in a hospital medical record is usually defined by state legislation. Additionally, accreditation organizations maintain standards regarding required documentation (e.g., the Joint Commission on the Accreditation of Healthcare Organizations). Therefore, although medical records from different hospitals in a given state may be compiled and stored differently, the required content is the same. This is useful to know, especially if documentation appears to be deficient.

Since the early 1990s, the 'traditional' medical record has been undergoing change. Today, it is not unusual for the content of medical records to be a combination of hard copy, electronic, and computerized formats. Therefore, surveillance staff should be aware that the hard copy medical record that is traditionally stored and managed by hospital medical records departments may not appear to be 'complete' with respect to documentation. Some documentation may be in computer files or on electronic storage files (e.g., CD-ROM, microfiche, microfilm).

A hospital medical record is generated for every admission and service encounter, and each record follows the guidelines for standard documentation. Some exceptions to this rule may apply in certain pregnancy outcomes. Programs should consult with hospitals and delivery sites for their procedures for outcomes other than live birth. The following are offered as possible scenarios:

- Live birth. The infant and mother will each have individual medical records.
- Live birth with neonatal demise shortly after birth. The infant may have a newborn medical record. However, most useful information will be in the mother's delivery medical record (e.g., if autopsy or cytogenetics laboratory work is done, the results may be placed in the mother's chart).

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- Fetal death. The fetus may have a medical record. However, most of the useful information will be in the mother's delivery medical record (e.g., if autopsy on cytogenics laboratory work is done, the results may be placed in the mother's chart).
- Elective termination. A medical record will be created only for the mother. Sometimes the admission at the hospital (or other site) will be as an outpatient.

There are other locations and places where births and other pregnancy outcomes can occur (e.g., in transit, in clinics, at home). Most, but not all, of these sites will generate a delivery medical record at least to fulfill federal and state requirements to complete a vital record. The depth of the information may be incomplete or inconclusive; therefore, additional investigative effort is usually required.

6.8.2 Type of Documentation in the Medical Record

Surveillance staff should be aware of the typical documents found in a medical record. This is true for staff conducting active case finding, as well as for staff conducting a medical records data quality audit for passive case ascertainment. Surveillance staff should consult with individual sites regarding records content requirements and how the documents are stored at the site (i.e., hard copy or computer file). The following are offered as examples:

- Face sheet. Contains demographic information, facility-specific information (e.g., medical record number, attending physicians, primary care provider, insurance).
- ➤ History and physical. Information is gathered and an exam is conducted at admission, at birth, and at various periods during the hospital stay (depending on the length of stay).
- Discharge summary. A document that is completed by a physician after a patient leaves the hospital. The summary pertains to a specific hospital stay and includes: admission diagnoses; pertinent medical history prior to the admission and problems, progress, and treatment during the hospital stay; a list of discharge diagnoses; and recommendations for follow-up, such as future visits to specialists and medications to be taken. At some hospitals a discharge summary may not be required for a very brief length of stay (e.g., less than 48 hours). Sometimes discharge summary information is recorded in the progress notes.
- > Consultations. Specialists such as neurologists, geneticists, or cardiologists also see the patient and provide diagnostic clarification.
- Progress notes. Health care providers (e.g., physicians and nurses) document treatment and plans.
- Diagnostic reports. Any procedure, whether invasive or non-invasive, requires documentation. This includes: diagnostic tests, laboratory analysis, surgery, cytogenetics, pathology, and autopsy. Sometimes, the final report will not be in the medical record (e.g., it may be in an electronic file or on file in a department of the respective site). Some results will be referred to in the discharge summary, progress notes, or consultation, while others may not be completed for several weeks (e.g., autopsy cytogenetics).

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6.8.3 Maternal Delivery Medical Record

In addition to standard documentation required in hospital medical records, the mother's delivery medical record contains unique pieces of information that are important for case ascertainment.

- Labor and delivery summary. Many hospitals use a standardized form to record important aspects of the outcome (e.g., time, weight, pregnancy risk factors).
- Prenatal medical records. Although the private obstetrician maintains these, some documents may be inserted in the mother's delivery record (or located in other places in the mother's hospital medical record). These include copies of the course of pregnancy management and results of prenatal diagnostic procedures, such as ultrasounds, amniocentesis, and cytogenetics analyses, particularly if a birth defect is detected prenatally.
- Pathology and laboratory reports. Pathological analysis is important in the case of fetal demise. Laboratory reports are important when there are suspected infectious disease or toxicology concerns in the mother. For example, there may be concerns about an exposure that could be passed along to the infant through breast milk.
- Autopsy. If an autopsy is performed on a fetal demise or neonatal death, the report is often inserted in the mother's medical record or may need to be tracked to the appropriate department.

6.8.4 Prenatal Medical Record

Currently, prenatal care may result in a woman having multiple medical records generated over the course of the pregnancy.

- ➤ Obstetrician's prenatal care medical record. This record contains documentation of how the pregnancy is managed. The content of this medical record is very similar to a hospital-based medical record; thus, it is important for birth defects surveillance. Sometimes the prenatal care medical record is inserted into the maternal delivery medical record.
 - *Prenatal care forms*. These are often in a standardized format and facilitate complete recording of information (e.g., laboratory work, family history, risk factors, genetic screens, and tests).
 - Flow charts of care. Prenatal visits, care and treatment, and patient discussions are documented, although often written by hand.
 - *Diagnostic tests*. The record may contain diagnostic tests, laboratory results, genetic counseling reports, consultations, and referrals to diagnostic centers.

Prenatal diagnosis is growing in importance for birth defects surveillance. There is a long history of chromosomal diagnoses that are detected prenatally through the procedures of amniocentesis and chorionic villus sampling. Many more diagnoses can now be detected through the use of high-level ultrasound. Technology and diagnostic methods will continue to advance in the area of prenatal diagnosis.

Referral prenatal diagnostics and diagnosticians. Referral centers specialize in high-risk pregnancy and have high-level diagnostic capabilities. Depending on the course of a high-risk pregnancy, the referral physician (diagnostician) may assume primary management of the pregnancy and may attend the delivery. However, usually, the referral diagnostic site and diagnostician do not follow the patient throughout the pregnancy. Medical records generated at the referral diagnostic sites may contain pertinent information from the primary obstetrician's

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office, including demographic information, index prenatal care history, medical history, risk factors, and reasons for referral. They also contain unique information for case ascertainment. Sometimes the referral prenatal diagnostics are inserted into the obstetrician's prenatal care medical record.

- *Diagnostic and laboratory results*. The medical record includes the results and discussion of the results.
- *Genetic counseling*. Documentation in this report includes significant family history, discussion of prenatal diagnosis, and discussion of prognosis.

6.8.5 Cytogenetic Laboratory Reports

Cytogenetic analysis may be performed at the hospital (in-house) or at freestanding laboratories. Programs are encouraged to use cytogenetic laboratories as data sources that consistently report cases. It is important for birth defects program staff to have some knowledge of basic genetics and the chromosomal terminology they are likely to encounter in medical records. For additional information on cytogenetics terminology (and corresponding abbreviations and symbols) refer to the reference manual, *International System for Human Cytogenetic Nomenclature (ISCN)* (Mitelman, 1995).

The report on cytogenetic findings is created by the lab that did the analysis. The report usually identifies:

- > Name of patient
- > Date of birth
- ➤ Referring facility and/or physician
- > Reason for referral (or suspected diagnosis)
- ➤ Result/karyotype
- ➤ Narrative regarding the analysis

Rarely does the report provide an address for the patient. This presents a challenge for a surveillance program that regularly receives case reports directly from the cytogenetic laboratory, since the laboratory may also perform analyses for patients from several states. Surveillance programs should develop quality control procedures that address this and other challenging issues when working with cytogenetic laboratories. One possible approach is to develop a list of the locations of the referring facilities and/or physicians.

The original report of the result of a cytogenetic analysis (or other test) is the property of the laboratory that performed the analysis. A copy of the report may or may not be sent to the referring facility or physician (or included in the referring facility's medical record). The results may be communicated orally or referenced in a medical record. The surveillance program should develop abstracting procedures for accepting a referenced cytogenetics analysis and for determining when it is necessary to locate the initial source of medical information.

There is a growing trend for hospitals to use out-of-state laboratories. Surveillance programs should investigate the feasibility, including legal authority, of using and contacting out-of-state laboratories.

6.8.6 Autopsy, Pathology, and Laboratory Reports

Pathology laboratories are usually associated with hospitals, while autopsies may be performed in selected hospitals or through coroner's offices. Autopsy and pathology reports are usually placed in the patient's medical record, but the autopsy report may be completed long after death (some states have 45-to 60-day time frames for completion of autopsies). Therefore, the autopsy report may not be filed with the admission medical record; it may be in the outpatient or 'other' section of the record. It is important to note that there are two completion status categories for autopsy findings or reports: provisional and final. Surveillance staff should place the highest level of diagnostic certainty on the final report.

Anatomical pathology laboratories usually produce high-quality case reports due to the exacting nature of the procedures performed during autopsy. An important exception to this is when the specimen is destroyed, macerated, or otherwise compromised, as is the case with many fetal deaths. When this happens, the autopsy and tissue analysis may be of limited value for birth defects case identification. Still, the autopsy report or tissue analysis will often provide the most definitive information on structural defects. Additionally, the type of tissue sample can provide useful information regarding the time frame of the pregnancy. Therefore, it is important to track and examine these reports.

Autopsy and pathology laboratories may have information management systems, manual or computerized, specific to the laboratory. Diagnostic information is usually accessible since these laboratories catalog their findings for forensic investigations, historical and legal archives, case studies, and medical board reviews.

Surveillance programs should understand that there might be varying degrees of quality in autopsy reports. Much depends on the expertise of a given pathologist or coroner, the majority of whom are not fetal and pediatric anatomical pathologists, the experts in this area. In some states these pathologists, and the hospitals or sites where they work, act as referral centers for specialized autopsies. Programs should consult with the respective pathologists and sites to better understand referral patterns in a given state and to evaluate the level of expertise available in this specialized area.

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6.9 Infant Risk Factors in Case Identification

A condition that affects an individual's chance of having a particular outcome is called a *risk factor*. Various maternal and pregnancy exposures and conditions have been associated with an increased risk for birth defects. Birth defects programs can use these risk factors to identify potential cases, either through including their ICD-9 codes on the discharge lists obtained from medical records departments, through reviewing logs for any entries citing these risk factors in addition to birth defects, or through identifying vital records with particular birth weights, etc.

However, even though certain factors are associated with increased birth defects risk, the majority of infants and fetuses with these risk factors will not have a birth defect. Thus, a large number of records will be reviewed that do not turn out to be cases.

Moreover, the list of risk factors that may be used as case-finding sources can become very large. It is possible that a large portion of the potential inclusion population will have at least one of the risk factors used as a case-finding source. Most risk factors only result in a small to moderate increase in birth defect risk, so the majority of records reviewed on this basis will not yield eligible cases. Such risk factor lists are developed from experience, logic, and research. Programs that use risk factors should evaluate the yield in their case identification approach and determine whether using risk factors as case-finding sources is useful to the program over time.

In the short term, the use of risk factors as screens for identifying potential cases of birth defects may be a valuable effort when the program is involved in a concentrated focus on a specific outcome, exposure, medical condition, or cluster investigation.

Surveillance staff may encounter various postnatal complications during the review of data sources and units. This information is most likely found in the infant's medical record, and often in progress summaries. In the situation of a fetal demise or stillbirth, the information is usually found in the maternal delivery chart.

The list below provides some examples of risk factors that may be useful as case-finding sources. Surveillance staff should use pediatric references to become familiar with newborn conditions and evaluate which conditions are appropriate to use for case finding. Passive case ascertainment programs should also evaluate the effectiveness of using risk factors. The majority of the items listed below are identified in data fields on the vital record (birth and fetal death, death certificates) and easily accessible to both active and passive case ascertainment surveillance systems.

Examples of infant risk factors include:

- ➤ Infants who weigh less than 2,500 grams (5 lbs, 8 oz) or are < 36 weeks gestational age
- Fetal and neonatal deaths
- Infants with a history of asphyxia at birth (Apgar score at 5 minutes less than 7)
- Infants admitted to neonatal intensive care or special care nurseries
- > Multiple births
- ➤ Infants with respiratory distress
- > Infants with heart murmurs

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Appendix 6.1 Data Source Described in Detail – Vital Records

Appendix 6.1 Data Source Described in Detail – Vital Records

Source or Site

- ➤ Birth certificates
- > Fetal death certificates
- ➤ Elective termination reports
- > Death certificates

Birth, death, and fetal death certificates provide a standardized way of reporting vital events that occur in a politically defined unit, a state. Vital records include facts about an individual and the specific circumstances regarding the reported event. Vital records are particularly important in that they fulfill two significant functions: they provide a mechanism for registering the occurrence of vital events, and they provide a mechanism for collecting demographic, social, and health information regarding the person in a standardized way. Integral to these functions is the fact that they are population based.

Legal or Professional Mandates

Federal law mandates birth and death registration. The lead federal agency is the National Center for Health Statistics (NCHS), Centers for Disease Control and Prevention (CDC). NCHS maintains the national birth and death registration system and is the recipient of vital records data from the states and territories. Recording births and deaths is the responsibility of the individual states and territories. The procedures and regulations regarding the reporting of these vital events are established by the individual states and territories. NCHS provides guidelines and recommendations for standardization of the information collected by birth and death certificates by promulgating standard certificates. Although federal law does not mandate the reporting of fetal deaths, there is an NCHS-recommended standard fetal death certificate. See http://www.cdc.gov/nchs for further information.

Mission or Objective

Provides a population-based statistical database of all births and deaths that occur in the United States.

Scope or Breadth

The birth, death, and fetal death certificates provide for registration of a defined vital event at a point in time. There are established criteria for what constitutes a live birth, but there is evidence to suggest that those criteria are not always followed. Registration of fetal deaths is usually defined on the basis of gestational age, with ≥ 20 weeks as the cut-off used by most states. Some states require the reporting of all fetal deaths, regardless of gestational age, and there is recognized underreporting of early fetal deaths.

Appendix 6.1 A.6.1-1 Case Ascertainment Methods

Operational Structure

- > Data. NCHS recommends standard data elements on birth and death (and fetal death) certificates. States are required to complete a minimum data set for national reporting and may add other data elements to their certificates. The birth certificate is usually revised and updated every decade. In 2003, the final drafts of a new version of the certificate are being reviewed. Please refer to http://www.cdc.gov/nchs for further information.
- > Certification. State statutes, regulations, and procedures stipulate who is responsible for certifying a live birth, death, or fetal death. The designated person is required to certify date, time, and place of birth/death as well as other priority areas on the certificate. The completion of death certificates has additional protocols, procedures, and instructions because of the many circumstances that may surround a death.
- Filing the certificate. State statutes, regulations, and procedures stipulate time requirements for filing. Although the timing varies among states, the certificate is usually filed with the state registrar's office within 5 to 10 days of the event. Many states now have methods for entering and filing birth certificates electronically. The timing for filing a fetal death certificate depends on state guidelines. Although filing a death certificate is required within a specified time period, it may not be complete at filing, as some data elements may be missing due to autopsy, coroner investigation, or other legal proceeding. These data may or may not be added subsequently and the certificate revised.
- ➤ Unique identification of an individual event. Each state has a numbering system that uniquely identifies the respective event.
- > Storing the information. Most states have a centralized database specifically designed to collect, amend, transmit, retrieve, sort, print, and analyze vital records information.
- > Reciprocity. Agreements with bordering states ensure reporting of life events occurring in neighboring states to the state of residence.

Types of Information Collected

- NCHS and other interested parties have developed a set of standardized data elements or minimum data variables that are required to be reported, as well as a set of recommended data variables and recommended standard certificates. Of importance is the unique identifying information per person, per event.
- The birth certificate and fetal death certificate are each divided into two sections: legal and statistical. The *legal section* contains the unique identifying information about the person, date, time, place, and type of life event. It is this portion of the certificate that registers the vital event. The information in the legal section is certified, and this is the part of the certificate that is issued to individuals when proof of the life event is required. The *statistical section* labeled "Information for medical and health purposes only" contains demographic, prenatal care, pregnancy risk factors, and medical conditions of the mother and of the newborn, including congenital anomalies. The statistical part is not released to the public, and many states do not keep the statistical part attached to the legal certificate. The statistical information is usually data entered and maintained in a database.
- ➤ The death certificate is a certified legal document, and it is available to authorized individuals in its entirety.

Appendix 6.1 A.6.1-2 Case Ascertainment Methods

Accessibility and Retrievability

States transmit vital records information to NCHS electronically. State laws and regulations stipulate how the information is made available for other users at the state level. Due to the confidentiality of the information, states protect the medical and health information on vital records from unwarranted or indiscriminate disclosure. Most states have legal safeguards in place to further protect the information.

- The information contained in the birth, death, and fetal death master index computer file is usually available to authorized public health programs. Sometimes confidentiality or security agreements are required.
- Many states copy the legal sections of the hard copy certificate into a permanent electronic storage format (e.g., microfiche, film, CD-ROM). The storage format is cataloged for easy information retrieval.

Strengths as a Data Source

- > *Timeliness*. Electronic filing allows information to be available to users as soon as the reports are filed in the state database. This may be as early as 30 days after the event.
- ➤ Population base. Provides statistical and denominator data.
- ➤ Unique identification. States assign a unique ID to each person, per vital event.
- Legality of case report. State laws require that some information must be certified for all births and deaths. Additional attention to legal procedures is required for death registration.
- Comprehensiveness as a data source. Over 97 percent of all births occur in a hospital or birthing facility. Out-of-hospital births are also registered because of the necessity for a child to have a birth certificate. There may be some underreporting of early infant deaths, and there is marked underreporting of fetal deaths at early gestational ages.
- Existing data set and one that is accessible over time. There is historical depth to vital records, but there have been major changes in format, content, and coding over time.
- > Record linkage. Useful in combination with other data for building the case record. The use of unique identifying information permits matching and linking with other data sources. Many states routinely link vital records to each other, for example a death certificate with the birth certificate, providing a linked birth-infant death file.
- ➤ Risk factor screening tool. Some data elements can be used to identify potential birth defects cases. Examples include: low birth weight, prematurity, low Apgar scores, neonatal death, multiple births.
- > *Intervention*. The availability of information in a timely manner is conducive to rapid intervention or investigation.

Weaknesses as a Data Source

- ➤ Data quality. Much of the medical information on the certificate has been shown not to be reliable.
- ➤ Case ascertainment. The birth certificate has been shown to underreport birth defects. As shown in Section 6.4, rates from this source are 1.5 percent, compared to 3 to 4 percent for hospital reporting and from using linked data sources.

Appendix 6.1 A.6.1-3 Case Ascertainment Methods

Liaisons and Partnerships

- ➤ Vital records/registrar's office. These are staff that are involved in managing the activities involved in filing the certificate. These staff often go to hospitals to train personnel in the procedures and methods of filling out the certificate. Other activities include amending a certificate, maintaining the centralized database, and cross-referencing other vital record certificates.
- ➤ Hospital. These are staff that are involved in providing information for completing the certificate. Includes medical records services, neonatal nursing, labor and delivery unit staff.

Hints and Tips

- Neonatal and infant death. A death certificate is issued upon death for any infant who was live born, regardless of duration of the pregnancy. These individuals will have a birth and a death certificate. There is no distinction in death certificates for 'neonatal' or 'infant' deaths. Many vital records divisions cross reference the birth and death certificate numbers to make sure that a birth certificate is issued if a neonatal or infant death is reported. Sometimes, the facility will overlook filing a birth certificate for an early neonatal death. Sometimes a fetal death certificate is filed as well as a birth certificate and/or a death certificate. In these situations further investigation should occur to determine the actual vital status at birth.
- The timing for filing birth and death certificates is similar. However, often the birth certificate is processed by vital records more quickly since many hospitals use the electronic birth certificate. It is important for birth defects programs to be aware of these timing issues if they refer children to services, especially if they refer children based on low birth weight, prematurity, and other severe conditions. Regardless of how quickly a case report is sent to the surveillance program, it is a wise practice to allow a period of time to elapse before referring a child with severe conditions. A time period to consider before referring a child to services is 60 to 90 days past the date of birth.
- Fetal death certificate. This certificate is usually issued for any pregnancy that results in a non-live outcome at the end of a pregnancy that is ≥ 20 weeks gestational age. What constitutes 'live' is subject to legal definition, and most states have clear guidelines in state statutes for what is considered a 'live birth'. Some states accept any sign of life (e.g., a pulse), regardless of the intent for the delivery (e.g., elective termination). Surveillance systems need to understand the definition of 'live birth' in their state. There may be instances when an Apgar score is a very low number (e.g., 1) at the first minute, and 0 for the fifth minute. Some states might count this as a live birth or a termination, depending on the age of the fetus and intent of the delivery. Some states have guidelines that exclude filing a fetal death certificate if the intent of the pregnancy delivery is for a termination, regardless of the gestational age.
- Termination reports. Some states collect statistical information on terminations. Often there is no identifying information; however, a birth defect may be listed as a reason for the termination. In most instances these reports do not have sufficient identifying information to link to an individual. Additionally, although some states require the filing of these reports, compliance is notably poor, such that there is an underreporting of these events and conditions.

Appendix 6.1 A.6.1-4 Case Ascertainment Methods

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Appendix 6.1 A6.1-5 Case Ascertainment Methods

Appendix 6.2

Data Source Described in Detail – Hospital Data Sets

Appendix 6.2 Data Source Described in Detail – Hospital Data Sets

Source or Site

- ➤ Hospital discharge data set
- ➤ Hospital admissions reporting system
- ➤ Hospital disease index

Discharge information is collected by the data source in a standardized format on individuals admitted for hospital-based services. This usually includes inpatient stays and outpatient surgery but may also include services performed in outpatient hospital clinics and emergency rooms.

Legal or Professional Mandates

- Federal law. The Health Information Portability and Accountability Act of 1996 (HIPAA) legislation defines electronic health care transactions, health information privacy and security standards, electronic signature codes, transaction standards and code sets, and unique health identifiers.
- ➤ Other professional mandates dovetail with federal requirements (e.g., Joint Commission on the Accreditation of Healthcare Organizations, American Hospital Association).

Mission or Objectives

Discharge data are collected for a wide range of possible uses. These include population-mix studies, market share analysis, hospital charges comparisons, length-of-stay studies, disease-specific and clinical information-specific case volumes, health care delivery access analysis, and crude and severity-adjusted death rate analysis. Discharge data are also used indirectly for financial analysis and billing.

Scope or Breadth

These data result from ongoing data collection and include all inpatient encounters. Some hospital data sets may also include outpatient encounters. The age of population served is defined by the mission of the site (e.g., a children's hospital may serve patients up to age 20 years). A discharge data set may consist of information from one hospital or may be a large statewide discharge data set of all hospitals. A record is created for each defined admission for hospital service. Discharge data sets are defined by a period of time (e.g., year) and are maintained so that they can be accessed over time.

Operational Structure

Information for the data set is collected from many places in the hospital, incorporated into the individual's medical record, and compiled in a standardized format. Health information management or medical records departments are responsible for processing the information that results in the data record for each patient encounter and in ensuring that the medical record contains the required documentation (content).

Appendix 6.2 A6.2-1 Case Ascertainment Methods

Type of Information Collected

Information included in this type of data set usually does not include patient names or Social Security numbers. The data elements collected, however, can lead one to a specific medical record. These data sets usually include: hospital identifier, patient medical record number, admission and discharge dates, patient type, patient date of birth, patient gender, patient's residential location (e.g., zip code, county), insurance source, charges, physician type, diagnosis and procedure codes in ICD format, and length of stay. Other information may be collected depending on the objectives of the data set.

Accessibility and Retrievability

Hospital discharge data sets are computerized and are used to generate routine reports and to respond to ad hoc queries. Some hospitals submit their discharge data to a larger organization that collects data from each hospital and compiles the information into a single statewide hospital discharge data set.

Strengths as a Data Source/Site

- Existing database. Data are easily accessible, retrievable, and available in a computerized format.
- > Specific information. Specific data fields can be identified and extracted from the data base.
- > Cross-referencing. Available data fields provide information that can be used to locate the medical record.
- ➤ Disease classification system. Information on discharge diagnoses and procedures is collected in a coded and standardized format, currently ICD-9-CM.
- > Timeliness. Data are usually available rapidly, within 6 months of discharge. Internet technology has increased accessibility and improved timeliness of data from this source for some states.
- Consistency of the data set. Data fields are filled in as required for billing and for federal reimbursements.
- > Follow-up. Hospitals have unique medical record numbers for patients, facilitating tracking and monitoring of cases.
- > Screening tool. Specific data fields, especially ICD-9-CM disease and procedure codes, can be selected for further investigation.

Weakness as a Data Source/Site

- Discharge set bias. The discharge data set is an administrative database. Information is collected and compiled using procedures that suit a particular health facility or meet other legal requirements. It is a services-, planning-, and financial-based data set.
- ➤ Population base. The service area and patient population for most hospitals are not well defined. Therefore, the relationship of the hospital's patients to a larger group of persons is difficult to quantify.
- > Disease classification system. Some disease categories and codes for birth defects are not specific and are limited in scope.
- Accuracy and clarity of diagnosis. Federal and professional standards are used to govern interpreting medical record documentation, which includes identifying a diagnosis and assigning a representative disease code. Suspected and rule-out conditions may be coded as a final

Appendix 6.2 A6.2-2 Case Ascertainment Methods

- diagnosis at discharge, leading to overreporting. A diagnosis may not be recorded for many reasons. Underreporting may occur if not all of the diagnoses documented in the patient's medical record are coded.
- ➤ Personal identifiers. Externally recognizable personal identifiers usually are not available. Data elements can be used to locate medical records. Some states have adopted legislation to permit the reporting of identifying information directly in the discharge data set for specific reportable conditions (e.g., Colorado adopted regulations to permit named reporting from hospital discharge data).
- ➤ Maternal information. Information on the mother is not recorded on the discharge data record for a newborn infant or child.
- No medical record is generated. In some circumstances a medical record is not created. For stillbirths and even some neonatal deaths, a medical record may not be created for the infant. Information pertaining to the delivery outcome, including autopsy and laboratory reports, will be in the mother's delivery medical record. However, the mother's chart cannot be coded to reflect an infant's medical conditions. Therefore, in these circumstances a birth defect diagnosis will be missed. Surveillance staff should use other data sources, such as the vital record, to identify a case where a medical record might not be created.

Liaisons and Partnerships

- ➤ Data processing unit. Hospital staff in a data processing unit manage the computerized information that is collected from various departments in the hospital. These persons can assist surveillance staff by accessing birth defects information that is stored in computer format.
- > State hospital associations. Some state hospital associations may serve the function of producing the statewide hospital discharge data set. They have a vested interest in providing customer service to a hospital by compiling aggregate statewide hospital data. Often these associations are also actively involved with the major users of the discharge data set (e.g., health departments, epidemiology programs, health planners).
- ➤ Health information management and medical records departments. The hospital's medical records staff are responsible for managing the information contained within a medical record. In addition to assembling the medical record and ensuring that it contains the required documentation, skilled personnel coders assign the disease classification codes and abstract pertinent information for administrative purposes (e.g., billing and the discharge data set). Since surveillance staff often use the disease classification codes to identify cases, it is helpful to maintain open communication with medical records departments regarding questions about hospital coding rules and other issues that might affect data quality.

Additional Comments

The hospital discharge data set is facing significant changes due to evolving federal regulations, including HIPAA and the conversion of the disease classification from the ICD-9-CM system to ICD-10-CM. HIPAA requirements address electronic transmission of data, standard data elements, and privacy and security issues. ICD-10 is a larger and more complex disease classification system, one that will affect the general taxonomy used for coding purposes.

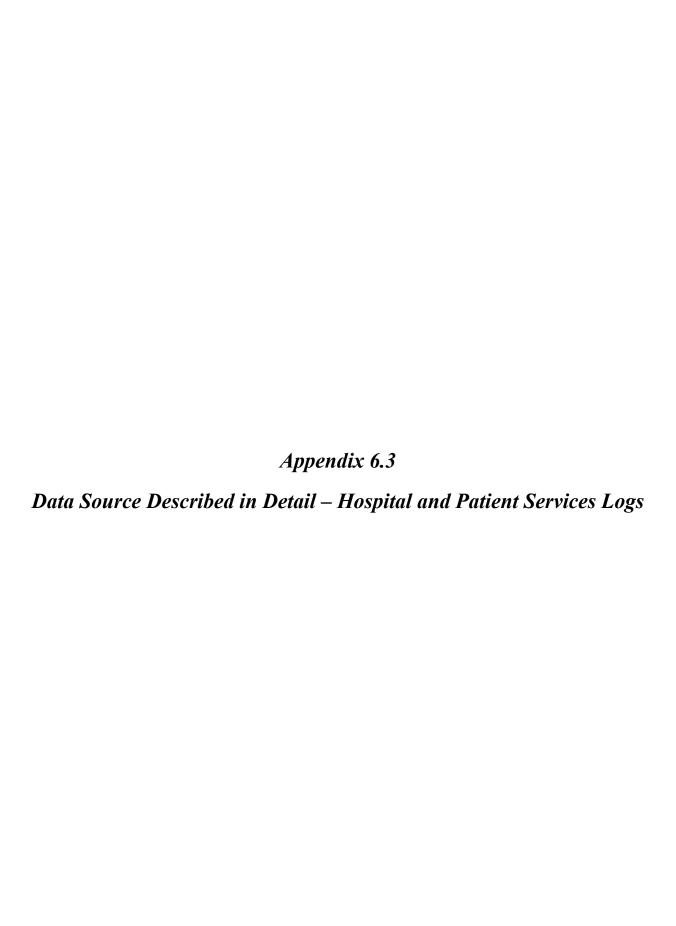
Appendix 6.2 A6.2-3 Case Ascertainment Methods

References

American Hospital Association (AHA). http://www.hospitalconnect.com/

Health Information Portability and Accountability Act of 1996.

Joint Commission on Accreditation of Healthcare Organizations (JCAHO). http://www.jcaho.org/



Appendix 6.3 Data Source Described in Detail – Hospital and Patient Services Logs

Source or Site

- ➤ Hospital unit logs
- > Patient services logs (in non-hospital settings)

Hospital units operate within a hospital or clinic and serve specific operational functions. Traditional units relevant to birth defects case ascertainment include Neonatal Intensive Care, Critical Cardiac Care, Labor and Delivery, and the Newborn Nursery. In some hospitals, units are their own departments, like Pathology and Surgery. A *unit log* is the documentation that provides information in general terms on the patients who used (or were admitted to) the unit.

Legal or Professional Mandates

- ➤ Legal state statute. Hospital-based unit logs are operated in accordance with hospital licensing and accreditation.
- ➤ Legal state statute. Non-hospital-based unit logs (e.g., birthing centers, prenatal diagnosis referral centers, genetics clinics), are usually operated in accordance with licensing guidelines.

Mission or Objective

Determined by site. Logs are used to record specific events or health system encounters in a particular hospital department or facility setting. Logs may also account for equipment use. The log represents an inventory of events or activities.

Scope or Breadth

Logs are point-in-time accounts of events. The unit log accounts for each entry or use of services into the specific area. Most logs identify an entrance time, and an exit time, as well as other information specific to unit requirements.

Operational Structure

Determined by site. Logs are designed to be read easily and to provide sufficient information to establish why the patient was in the unit or department.

Type of Information Collected

Determined by the site. Generally, logs are used by surveillance programs as a case identification screening tool. Most logs provide enough cross-referencing information to support follow-through or tracking. This includes name, date of birth, medical record or other identification number, and current date and time. Additionally, information is collected specific to the purpose of the encounter. Examples include:

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- Labor and delivery log. Prenatal information, maternal risk issues, prenatal diagnosis, and event or outcome measurements.
- ➤ Neonatal Intensive Care Unit (NICU) log. Event/outcome measurements, perinatal medical issues, diagnosis, other risk factors.
- > Surgery log. Preoperative diagnosis, possible risk factors.
- > Prenatal diagnostic center log. Prenatal information, referring physician, referring diagnosis, procedure, medical risk factors.

Accessibility and Retrievability

Logs are used as management tools within individual facility units. Therefore, information is gathered for and used by the unit and, possibly, by the facility. While some information may be collected and entered into a database, most logs consist of paper copy record books or reports.

Strengths as a Data Source

- Timeliness. Information is recorded in real time, as events occur. Rapid identification of potential cases is possible.
- > Consistency in recording information. The population base is well defined for each particular unit since each service encounter is recorded. For example, if a surgical procedure was performed at the site, a surgical log will record the episode.
- > Case identification screening tool. Generally, enough information is recorded so that surveillance staff can identify potential cases for further investigation.

Weaknesses as a Data Source

- Effort to retrieve the information. Generally, logs are kept in hard copy format and are based on a handwritten recording of events. Review of the information can be effort intensive.
- Accuracy and clarity of clinical information. Information recorded may be inaccurate or incomplete with respect to diagnoses or medical conditions. For example, a prenatal ultrasound log may state 'referred for cardiac irregularity'.
- Documentation in the log. Information recorded on a log may be of limited use for case identification. Sites establish criteria for log documentation to meet internal or ward management objectives, not for disease coding. As such, the information is most relevant for immediate patient management rather than as a tool in medical diagnosis and treatment.
- ➤ Different logs within the data source may provide conflicting information on the same patient. Surveillance staff should develop management tools to keep track of information recorded from different logs.

Liaisons and Partnerships

➤ Unit staff. These persons are usually front-line staff who work in the unit and have a use for the information that is recorded.

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> Office staff. These are the persons at the unit who are usually responsible for compiling statistics for the unit and who monitor occupancy. They may be able to assist the surveillance staff in identifying efficient ways to access log information. For example, they may be able to generate a computer listing of the log or provide a photocopy of the log sheet.

Issues to Consider

Surveillance program time and efficiency issues. Unit logs usually require surveillance staff to spend time identifying potential cases on the log and following up by reviewing medical records. Case identification screening criteria and the quality of information included in a log are significant factors to consider when evaluating the amount of time spent on finding cases using this source. Inefficiencies result when follow-up medical records reviews result in too many non-cases. Time and effort evaluations should be conducted for the case identification processes involved in using unit logs.

Unit logs serve as a management tool for individual components of a facility. Therefore, a potential birth defects case may show up on multiple logs. It is useful to compare the information recorded at each unit within the data source and to develop a surveillance management tool that tracks case-finding activity. Such a tool will minimize staff time spent requesting and reviewing a medical record multiple times.

References

None.

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Appendix 6.4

Data Source Described in Detail – Genetic Services

Appendix 6.4 Data Source Described in Detail – Genetic Services

Source or Site

- ➤ Regional/state genetics networks
- ➤ Hospital-based genetics clinics
- University-based genetics clinics
- Provider-based genetics clinics

Geneticists and dysmorphologists are skilled at evaluating a constellation of findings, providing differential diagnoses, and determining the definite medical condition. They use diagnostic procedures such as chromosomal analysis and genetic testing, as well as drawing from their personal experiences and extensive literature in evaluating a patient.

The information from this data source is of high quality.

Legal or Professional Mandates

- Legal. State statutes for hospital-based clinics. These are operated in accordance with hospital licensing and accreditation.
- ➤ Legal contract. Specified in individual contracts or collaborative agreements.
- ➤ *Professional*. Certification and professional credential as required.

Mission or Objective

Genetic diagnostic and counseling services, therapeutic management of genetic diseases.

Scope or Breadth

Clinics may include prenatal, pediatric, and/or general population. Some may be specialized by disease category (e.g., Down syndrome, cystic fibrosis). Some providers include diagnostic and research laboratories, clinical research centers, and off-site clinics.

Operational Structure

Genetics clinics may be set up as a referral site (i.e., to provide a diagnosis back to the referring physician), for services (i.e., for ongoing treatment and consultation), or for research or study (i.e., database).

Type of Information Collected

Depends on the focus of the encounter (i.e., prenatal, pediatric, and counseling). As a rule, genetics clinics collect a core set of information for each patient, including demographic data and family medical history. A detailed physical exam and diagnosis, if known, as well as a case summary, is also usually available.

Appendix 6.4 A6.4-1 Case Ascertainment Methods

Copies of outpatient diagnostic tests and procedures may also be found. Clinics may use multiple disease classification systems depending on the diagnosis (e.g., ICD-9-CM, ISCN or *International System for Human Cytogenetic Nomenclature*, [Mitelman, 1995]) and/or use proprietary coding systems (e.g., POSSUM, Mendelian Inheritance in Man). Clinic charts may also include letters and notes from other physicians, results of research studies, or diagnostic testing that borders on research.

Accessibility and Retrievability

Usually the medical charts for clients/patients are available at the clinic site for review and abstraction. Many clinics collect information in database format for insurance purposes, clinic needs, and networkwide data collection. Due to the nature of the information gathered, the data often are retained permanently. However, state statutes should be consulted for statute of limitations for health information.

Strengths as a Data Source

- Accuracy. High quality. The status of a diagnosis is qualified (i.e., the definite, rule out, possible). Although some patients never get a definitive diagnosis, the differential diagnosis is usually provided.
- Level of detail. High quality. Specific information on syndromes (identification and description of dysmorphic features) and chromosomal anomalies is often provided.
- Case identification. Specialty clinics, like those for genetics, are important outpatient data sources. Previously unknown cases may be identified for the surveillance program.
- > Case identification or screening. This is a useful source for prenatal diagnosis cases. Clinics may provide diagnosis and/or genetic counseling services.
- Retrievability. Most pertinent information is entered into an electronic file (i.e., a database). This facilitates requesting specific pieces of information that can be extracted in electronic format.

Weaknesses as a Data Source

- ➤ Population base. May not be well defined.
- Incomplete information. Nature of the clinic business or the clinic encounter determines whether the complete diagnostic picture is available (i.e., the case may be referred for cytogenetics laboratory confirmation only).
- Timeliness of diagnosis. Some diagnoses are not confirmed until multiple diagnostic procedures have been conducted. Some syndromes take a long time to be diagnosed definitively.
- Follow-up. Often a case is referred for consultation and is lost to future tracking. This is important if the diagnosis is reported to the surveillance program as possible or rule out and is in the continuing or discovery phase.

Appendix 6.4 A6.4-2 Case Ascertainment Methods

Liaisons and Partnerships

- ➤ Genetic counselors. Clinics are often staffed by genetic counselors who contribute documentation concerning a patient's evaluation. They are often accessible to surveillance staff if a medical records review or other follow-up is needed.
- ➤ Database managers and other office administrators at clinic sites. Clinical information is often abstracted from documentation in the medical record for billing, research, or other clinic use. These persons can assist the surveillance staff in identifying efficient reporting and case identification methods.
- Network system managers. Regional genetics information may be collected and compiled in a database. Like hospital discharge data, regional genetics information is collected from participating clinics in a standardized format and compiled in a centralized format. Surveillance staff can utilize the efficiency of accessing a centralized database and bypass having to collect the case reports from individual clinics. Of importance is the fact that data from these sources are unlikely to include personal identifiers

Issues to Consider

- > Scope of information collected. Genetics clinics may collect information and provide a diagnosis that extends beyond the types of defects included in a birth defects surveillance system. Passive case ascertainment systems should be precise in specifying the diagnoses that are included in the program's case definition and which are reportable. Active case ascertainment programs could improve efficiencies by developing a more precise list of diagnoses and medical conditions that can be used to screen for potential birth defects cases in the database or log of the clinic.
- Confidentiality issues. Genetics information may be protected by additional federal or state statutes. The surveillance system should research applicable legislation, and if necessary, strengthen security procedures and processes in the surveillance system.

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Appendix 6.4 A6.4-3 Case Ascertainment Methods

Chapter 7 Data Quality Management

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7.1 Introduction

The credibility of a birth defects surveillance program is built on a foundation of high-quality data. Information and results that are derived from surveillance data should be accurate, complete, and timely. Data quality influences the results of descriptive epidemiologic studies and, therefore, their interpretation. Data quality also affects the extent to which information can be utilized for planning, prevention, and intervention.

In this chapter, we will discuss some of the issues that affect the quality of data in surveillance systems and suggest methods for quality improvement. In Section 7.2 we present criteria designed to produce high-quality data. In Section 7.3 we introduce some relevant terminology. In Section 7.4 we discuss the relationship between data sources and quality, and in Section 7.5 we outline the distinctions between timeliness on the one hand versus thoroughness and completeness on the other. Sections 7.6 and 7.7 present various aspects of quality control and quality assurance, stressing the differences between the two. The importance of computer technology in support of quality improvement is particularly highlighted in Section 7.7. Nine specific quality improvements methods are discussed in detail in Section 7.8. References cited in this chapter may be found in Section 7.9.

This chapter contains a Data Sources Descriptive Assessment Tool that may help surveillance staff systematically evaluate the various data sources available to them (Appendix 7.1).

Chapter 7 7-1 Data Quality

7.2 Criteria for High-Quality Data

High-quality data have a positive cascading affect on a surveillance program's outcome measurements – such as accuracy, completeness, and timeliness – which, in turn, can be monitored as a means to improve program performance. The term *quality* has many definitions and interpretations depending on use and intent. Philip Crosby, a total quality management expert, defines quality as "the conformance to agreed and fully understood requirements" (Dale and Bunney, 1999). In the surveillance field, this translates into the identification of a target, bench mark, or goal that defines the requirements against which results are measured.

Some experts in surveillance have suggested that the most important measurement indicators (or criteria) related to high-quality data are described by the mnemonic TACOMA (NAACCR, 2000). Data must be Timely, Accurate, Complete, Oriented, Measurable, and Applicable. The relative importance of these factors should be weighed and balanced, individually and in total, against the program's objectives and resources.

In the next section we define the terms on which the TACOMA mnemonic is based – timeliness, accuracy, completeness, oriented, measurability, and applicability – as well as several additional terms important for an understanding of data quality issues.

Chapter 7 7-2 Data Quality

7.3 Terminology

Timeliness The extent to which data are rapid, prompt, and responsive. For example, a birth

> defect case should be ascertained or reported to the program shortly after diagnosis. With rapid case identification, the program is able to provide timely prevention and intervention services, respond quickly to investigations, and

monitor trends.

Accuracy The extent to which data are exact, correct, and valid. For example, accurate

> diagnostic data affect a program's ability to provide reliable disease rates and to maintain data comparable to those from other programs. Diagnostic accuracy reflects the program's standard to conform to agreed-upon case definitions and

requirements.

The extent to which data are all-inclusive and comprehensive. For example, are all **Completeness**

of the cases of birth defects that occur within the target population, within a

specified time period, identified by the surveillance system?

The extent to which data are focused, targeted, and intended. For example,

programs should collect only those data that are appropriate to their goals and objectives. Programs should determine which data variables should be collected, how quickly they can be collected, and the resources available to be devoted to their collection. Having an oriented perspective parallels the 'minimum necessary' privacy standard of the Health Insurance Portability and Accountability Act or HIPAA (i.e., identify and use only what is necessary). (See Chapter 2 on

Legislation for additional information on HIPAA and Chapter 4 on Data Variables

recommended for consideration by birth defects surveillance programs.)

Measurability The extent to which data are quantifiable, calculable, and objective. For example,

the conformance to agreed-upon data definitions provides the foundation for

quantitative evaluations.

The extent to which information is relevant. Outcome measurements should be

designed to promote data utilization. Information derived from the data should be

beneficial to the target population or to public health interests.

Comparability The extent to which the data in one data set conform with those in other data sets.

For example, programs that agree to adhere to standard data definitions and case definitions produce data that can be evaluated and weighed against one another.

The extent to which data collection activities are meticulous and exhaustive in **Thoroughness**

completing a case abstract or case record. In other words, each data field on case

abstracts and case records should be filled in.

Strategically planned results that may be quantitative or qualitative. Criteria, such as those described by TACOMA, or other defined factors, are specifically selected measurements

(and developed) to evaluate, track, and monitor a program target, goal, or

benchmark. Desired outcome measurements are often developed in the planning stages of a surveillance program, for performance evaluations, and when adding new projects. Staff should identify the type or category of results to be measured in order to evaluate progress in achieving program goals and objectives (or study

objectives, project targets, etc.).

Oriented

Applicability

Outcome

7.4 Data Sources and Quality

Depending on the case ascertainment approach, birth defects cases are found-at or reported-from data sources. Therefore, the importance of the role data sources play in case ascertainment and surveillance should not be underestimated.

Quality issues surface because of variations among data sources. Some data sources may provide diagnostic information, but may lack important demographic information. Some may be service-focused, such that a precise diagnosis may not be important. Others may provide in-depth information on a specialty area, but may not identify other conditions that co-occur. Still others are administrative databases.

A single data source has the potential to affect multiple outcome measurements. For these reasons, programs should evaluate each data source in order to describe its basic characteristics, as well as to identify its potential strengths and weaknesses. A descriptive assessment tool should be designed to answer specific questions about each data source in relation to surveillance requirements. An example of such a tool is provided in Appendix 7.1 (Data Sources Descriptive Assessment Tool).

Quantitative evaluations should include outcome measurements for accuracy, completeness, and timeliness. Often data sources are evaluated in combination with other quality assessments. For example, diagnostic accuracy may be evaluated by staff reviewing a medical record to confirm a diagnosis that was identified-at or reported-from a data source. In this example, the data source is part of the evaluation because it is where the diagnosis case report originates; however, other aspects of the case ascertainment process may be evaluated as well.

Examples of quantitative evaluations are provided in Section 7.8 of this chapter on Quality Improvement Methods.

The program should:

- > Use the data quality criteria in TACOMA as a guide when identifying outcome measurements and when evaluating data sources.
- ➤ **Identify** other factors that are important to consider, including those that relate to staff resources, such as 'location of site' and 'volume of case reports' (in relation to distance traveled).

7.5 Timeliness Versus Thoroughness and Completeness

Surveillance systems generally have limited resources to use in meeting program objectives. Additionally, staff face dilemmas in terms of prioritizing resources to achieve the outcome measurements of timeliness, thoroughness, and completeness. Should a program set a goal of timely data at the risk of potentially missing cases? Or risk losing timeliness by setting a goal of the most complete surveillance database? It is important for programs to achieve a balance that suits their needs, while also being responsive to external requirements, such as guidelines for submitting data to the National Birth Defects Prevention Network (NBDPN), as discussed in Chapter 10 on Data Collaboration and Dissemination.

Timeliness improves a system's ability to be responsive for investigations, up-to-date for monitoring trends, and current for referral to services. Thoroughness is a measure of finished versus unfinished case abstracts and case records. Clearly, data fields that are empty or inconclusive are not useful for most outcome measurements. Completeness is important because descriptive epidemiology – including the calculation of birth defects rates – is more comparable, accurate, and reliable when a surveillance program is confident that all cases have been ascertained.

When prioritizing resources to balance the quality indicators of timeliness, thoroughness, and completeness an important outcome measurement recommended by NBDPN is that the surveillance database be 95% complete by two years past the date of birth or fetal demise. Some programs may have a longer time period for reporting birth defects and, therefore, have a longer time period for case ascertainment. Still, it is important that surveillance systems be sufficiently responsive so that complete and timely data can be turned into useful information.

Programs should evaluate the factors that impact timeliness, thoroughness, and completeness. Often resources can be used more efficiently and effectively by streamlining or redeveloping procedures in individual areas, such as case finding, data collection, and data processing (see Section 7.8 on Quality Improvement Methods).

Timeliness, thoroughness, and completeness are often intertwined and affect other quality assessments. For example, the quality control methods that evaluate case finding and case abstracting may include outcome measurements for timeliness and thoroughness. Data source evaluations include a timeliness measurement.

The program should:

- **Develop** productivity guidelines and standards.
- ➤ Use TACOMA criteria, especially 'oriented' and 'applicable', to assess the factors that challenge timeliness and completeness.
- ➤ Use computer technology to improve timeliness. For example, consider using the Internet for case reporting. Internet and electronic reporting also ease the burden of case reporting at data sources.
- **➤ Monitor** timeliness.

7.6 Quality Control and Quality Assurance

'Quality control' (QC) and 'quality assurance' (QA) can be defined as a set of methods, activities, and procedures designed to improve the results of specific outcomes. For birth defects surveillance programs, these outcomes are related directly to surveillance functions, such as case ascertainment and data collection. Although active and passive case ascertainment systems may use different methods and procedures for improving data quality, the goal is the same, namely high-quality data.

Quality control is a retrospective and reactive approach to improvement that focuses on discovery and detection. Deficiencies and inaccuracies are found, resolved, and fixed so that final results or outcome measurements are accurate. As a result of QC procedures, high-quality data are created at the back end. In QC, the emphasis is on checking, investigating, containing, and adjusting (Dale and Bunney, 1999).

QC procedures may include re-case finding, re-abstracting, validity audits, timeliness monitoring, and data source evaluations. QC can also be used with data linkage, especially as this involves checking selected data fields, including birth weight, date of birth, name, etc. The results of QC procedures are used to evaluate, adjust, or correct the original data that were collected or the original circumstance that occurred

Quality assurance is a proactive approach to improvement that focuses on prevention. Program functions are designed and activities are planned in advance to avoid inaccurate or deficient data. As a result of QA procedures, high-quality data are created at the front end or design stage. Often, the results of a QC method lead to QA activity. The QC method detects a deficiency, and the QA method redesigns the process to prevent its recurrence (Dale and Bunney, 1999).

QA procedures may include documentation (e.g., case finding, abstraction, medical records review, disease coding, data entry), the use of selective data sources, and the development and maintenance of the database infrastructure. Additionally, QA procedures can be implemented when specific outcome measurements require consistently high-quality data. Examples include (1) using an expert clinical reviewer to routinely evaluate case abstracts for data accuracy and thoroughness and (2) conducting medical records reviews to confirm a diagnosis prior to the data being used for projects like rapid case ascertainment, investigative inquiries, or statistical monitoring of trends. QA is cost efficient in the long run. Finding and solving problems can be time consuming and resource intensive, and unless the process is fixed, the same problems will continue to recur.

Maintaining high-quality data requires continual attention to improvement. Program performance is enhanced when quality improvement procedures are integrated into program operations and conducted in a consistent and systematic manner.

Refer to Section 7.8 (Quality Improvement Methods) in this chapter for specific examples of quality control and quality assurance applications.

The program should:

- ➤ Maintain documentation on program procedures, especially as these affect case ascertainment and data collection activities.
- **Record** and date decision items.

- ➤ **Identify** the sources of potential data quality issues and prioritize the impact of each on case ascertainment and surveillance. Some situations are provided in the 'quality issues' sections in other chapters in these guidelines.
- ➤ Use the TACOMA quality indicators to develop outcome measurements for evaluations. Of particular importance are quantitative evaluations of accuracy, completeness, and timeliness.
- ➤ **Design** meaningful evaluations, develop benchmarks, and track improvements. Quality assessments should be used to guide any decision to change or modify the program's practices and procedures.
- ➤ Use the results of quality control to design quality assurance procedures. Quality assurance is a self-propelling mechanism that ensures continual quality improvement.

Chapter 7 7-7 Data Quality

7.7 Quality Control and Quality Assurance in the Surveillance Database

Computer technology provides many opportunities to implement quality control and quality assurance procedures. Computerization can promote standardization, perform queries on selected criteria, monitor timeliness, reduce duplication, and generate reports.

Quality assurance can be built into the design, development, maintenance, and expansion of the surveillance database. It is essential that the computer system address, at a minimum, the requirements of case ascertainment and data collection, data entry, information management, and statistical analysis. The system must also ensure security and privacy for the health information that is stored electronically (see Chapter 9 on Data Management and Security).

A database system should be documented thoroughly, with methods in place to track changes in procedures and processes and to identify security safeguards.

Standardization of data variables is an important quality assurance procedure. Data fields should have discrete definitions, and programs should standardize the information in a data field with unique codes or pre-formatted text. Drop-down windows can assist with this by providing choices and by placing limits on the options for the data field. Drop-down windows also prevent keying errors during data entry. Data fields can be programmed to perform logic checks for dates, time, age, gender-specific disease codes, and geographic information. Calculations can be programmed into data fields for measurements (such as weight, height, and head circumference) or can be programmed to complete a 'missing' measurement for a data field.

Software technology can also provide excellent resources for quality control. Procedures can be developed to monitor timeliness, productivity, and progress. Transaction logs can be used to monitor key activities and tasks. A posting-date field can be used to track staff entries as the case ascertainment process proceeds. Posting fields can also be used to monitor data source reporting trends, data collection activities, and data processing functions.

Any number of outcome measurements can be developed to track quality indicators, including measuring accuracy and completeness. Additionally, computer technology is uniquely suited to detect duplicate cases in the surveillance system. Information can be cross-linked on many different data fields, including name, date of birth, hospital of birth, mother's maiden name, etc.

There are almost limitless ways that computer technology can be used in quality control. The database integrates and supports surveillance activities. As such, the inputs and outputs of the database play a role in each TACOMA quality indicator. A well-designed database improves program efficiencies, outcome measurements, and data utilization (see Section 7.8 on Quality Improvement Methods).

The program should:

> Identify situations in case ascertainment and data collection where computer technology can be used to detect or prevent problems and to track measurements.

Chapter 7 7-8 Data Quality

7.8 Quality Improvement Methods

Methods to measure and ensure high-quality data may vary depending on the approach to case ascertainment.

In active case ascertainment, field staff engage in the process of case identification, including gathering information and confirming a diagnosis for the case abstract. Quality control is directed at improving the way staff ascertain cases. In passive case ascertainment, the surveillance system receives case reports from data sources. Staff are not engaged in collecting the information on a case report. Additionally, a diagnosis reported on a case report is not usually confirmed prior to entry into the database. Therefore, in passive case ascertainment, quality control is directed at improving the results of the data collection process.

Although the ascertainment approaches are different, quality control and quality assurance methods can be used to achieve comparable levels of data quality across surveillance programs regardless of the ascertainment approach used.

While the list is not all inclusive, some of the methods used most frequently by birth defects surveillance programs for quality control are described below. Some are useful regardless of the case ascertainment approach and can be modified to suit the specific programmatic needs.

On the following pages we describe the following quality improvement methods in detail:

- Re-case finding
- > Re-abstracting
- > Validity audits and medical records reviews
- Clinical review
- ➤ Reliability and inter-rater agreement checks
- > Timeliness measurements
- > Data source evaluation
- ➤ Comparison/verification between multiple data sources
- Computer technology

Improving Quality through Re-Case Finding

Purpose

To evaluate the accuracy and comprehensiveness of the case-finding process.

Background

The case-finding process, used primarily in active case ascertainment, involves staff identifying potential birth defects cases at data sources.

Method

For re-case finding, perform the same steps and functions as for case finding. Develop procedures to evaluate results from the different pathways and steps in the process. Re-case finding should be conducted on a sample of information sources. The sample should consist of an appropriate number of entries, either from a single log or from multiple logs.

Outcome Measurements

- Evaluation of results between the original case-finding activity and the quality control process. This includes calculating the false positive and false negative rates at different steps in the case-finding process. In other words, this QC procedure evaluates the decision making that results in identifying a case versus a non-case.
 - Compare the QC list and the original staff review list of potential cases found at a data source during initial case finding. This is the list that identifies which cases go on to a medical records review and which do not.
 - Compare the results of re-reviewing the medical records. This involves
 QC re-reviewing medical records that were selected for review during the
 original case-finding activity and reviewing (for the first time) some
 medical records that were not on the original staff review list.
 - Determine the timeliness of the case-finding process.
- Evaluation of compliance with case-finding procedures, including assessing decision-making skills.

Frequency

It is important to develop a benchmark for re-case finding and to monitor outcome measurements periodically. The frequency with which re-case finding is conducted should be based on the demonstrated expertise and proficiency of the staff.

Quality Assurance

- Update case-finding procedures.
- Streamline the process to improve timeliness.

Tips

The case-finding process is a critical step in case identification. Not only is it important to evaluate staff effectiveness in identifying cases (and not missing any), it is also recommended that programs evaluate program efficiencies in case finding. For example, programs should evaluate the types of conditions that are considered potential cases. An evaluation might consist of determining how many confirmed diagnoses resulted from using a 'potential condition' in the initial steps of case finding. Some programs include ICD codes (i.e., searching through a hospital's disease index) as 'potential conditions'. An evaluation might consist of evaluating the effectiveness of searching using disease codes to identify a potential case in relation to whether specific codes were predictive in identifying a true birth defects case (i.e., an abstract is created).

Improving Quality through Re-abstracting

Purpose

To evaluate the accuracy and comprehensiveness of information that is entered on a case abstract form (hard copy or computer screen).

Background

Abstracting, used in active and passive case ascertainment, is the process of gathering and recording specific information from logs, medical records, or other information sources onto standard case abstract forms or computer screens.

Method

For re-abstracting, gather and abstract information from the same information source and record the data using the same abstract format (e.g., hard copy or computer screen). Re-abstracting should be conducted on a sample of information sources and a range of diagnosis categories

Outcome Measurements

- Comparison of the results of the quality control method to the results from the original case abstract and evaluation of the differences. Evaluation of the percentage and type of false positive cases.
- Identification of types and categories of errors or deficiencies. This may include disease coding, incomplete or missing information, and data entry errors. Includes the types of data variables that are problematic.
- Evaluation of compliance with abstracting procedures and guidelines.
- Determination of the timeliness of the abstracting process.

Frequency

It is important to develop a benchmark for re-abstracting and to monitor outcome measurements periodically. The frequency with which re-abstracting is conducted should be based on the expertise and proficiency demonstrated by staff.

Quality Assurance

- Update case abstracting guidelines.
- Provide training in disease coding, as applicable.
- Incorporate additional standardization into the data entry process. For example, provide drop-down windows to select and limit choices and to prevent key stroke errors.

Tips

Conduct an *abstraction form review* to identify differences and errors on completed abstraction forms. The abstraction forms should be checked for completeness, logic, and correct coding. Additionally, it is useful to categorize the types of data variables that are problematic to abstractors. For a given time period, QC should document, for each field staff member, the total number of abstraction forms reviewed and the number that have errors, such as incomplete or illogical data and incorrect coding.

Chapter 7 7-11 Data Quality

Improving Quality through Validity Audits and Medical Records Reviews

Purpose

To evaluate the accuracy and comprehensiveness of a diagnosis that is reported by a data source or represented in a listing (e.g., hospital disease index) at a data source.

Background

In programs using passive case ascertainment, birth defect cases reported by data sources are accepted without confirmation. Active case ascertainment systems may use a listing of diseases provided by data sources, in disease-coded format, as part of case finding.

Method

The medical record, or other medical information report, is reviewed at the site or data source that reported the diagnosis or provided the diagnosis in a listing. This method is also used in the data sources audit.

Outcome Measurements

- Predictive validity. This is the degree to which an original measurement (e.g., reported diagnosis) successfully predicts a valid or confirmed outcome of interest. In other words, it represents agreement between the case report from the data source and the medical records review performed by surveillance staff.
- Evaluation of missed diagnoses. In other words, how many more diagnoses were identified by the medical records review process.
- Identification of disease-coding issues, especially as this pertains to data sources that report birth defects in a coded format (e.g., administrative databases such as the hospital discharge data set).
- *Incorporation of an evaluation of the data source* with the validity audit.
- *Timeliness* of the review process.

Frequency

Passive case ascertainment systems rarely have the resources to confirm all reported cases through medical records review. Therefore, the frequency of validity audits depends on program resources, requirements, and priorities. However, it is important to develop and maintain a certain level of validity audits. Programs should develop benchmarks, set goals, monitor results, and adjust program procedures.

Quality Assurance

- Identify and use data sources that report a confirmed diagnosis.
- Select diagnoses for consistent, concurrent, and timely validity audits. It is recommended that the diagnoses be from the set of birth defects that are reported to NBDPN. This QA procedure is primarily for passive case ascertainment systems.
- Identify disease codes that are problematic for describing birth defects precisely. Prioritize which ones should have a consistent validity audit. This QA procedure is applicable for passive case ascertainment systems that use the ICD-9-CM coding system, and can be adapted to accommodate active case ascertainment programs that use the hospital disease index during case finding.

Improving Quality through Validity Audits and Medical Records Reviews (continued)

Tips

Validity checks are a quality control tool. Although used primarily by passive case ascertainment systems, the tool is relevant for active ascertainment programs as well (e.g., active ascertainment key data entry systems or on-line abstracting). Validity checks in birth defects surveillance provide a way of evaluating the accuracy of what was reported (or represented) compared with what was 'validated' or confirmed after an investigation or medical records review.

Improving Quality through Clinical Review

Purpose

To review the diagnoses listed on the case abstract or in the case record for accuracy and plausibility.

Background

Information on birth defects cases is gathered and compiled by staff in active case ascertainment. Information is reported and collected from data sources in passive case ascertainment.

Method

Case abstracts or case records are examined by a designated clinical expert.

Outcome Measurements

- Evaluation of the information recorded on the case abstract.
- Identification of abstracting or coding problems.
- Percentage and types of agreement or disagreement with clinical review result

Frequency

Programs should develop a benchmark for volume and types of case abstracts that should be reviewed and monitor the rate of agreement. In other words, the program should determine whether all case abstracts should be reviewed, or merely a percentage.

Quality Assurance

- Update and standardize abstracting and disease coding procedures.
- Train staff in the deficiencies cited and evaluate compliance concurrently.
- Increase the volume of clinical reviews, as required.

Tips

A clinical reviewer should be proficient at disease coding since the literal text of the diagnosis needs to be translated into the most accurate disease code.

In passive case ascertainment, the medical records from all data sources that reported a diagnosis for a respective birth defect case should be available to the clinical reviewer. Document the policies and procedures for the clinical review to ensure standardization. Include instructions for assigning the disease code.

Chapter 7 7-14 Data Quality

Improving Quality through Reliability and Inter-Rater Agreement Checks

Purpose

To evaluate rate of agreement between two or more persons for the outcomes of interest.

Background

Results of case ascertainment and data collection should be consistent, especially when staff are required to make abstracting decisions.

Method

- Dual-entry coding system (double-checking of assigned code). At least two coders assign codes from the same list of diagnoses.
- *Dual-entry data entry*. At least two staff key information from the same case abstract into the surveillance database.
- *Dual clinical review*. At least two clinical reviewers examine the same abstracts and provide results.
- Dual medical records reviews. At least two staff review the same medical records and abstract information per program procedures. This may include evaluating disease code assignments. Some passive case ascertainment programs may benefit by including a clinical expert in this inter-rater reliability evaluation.

Outcome Measurements

- Rate of agreement
- Type of deficiencies
- Compliance with abstracting and other program procedures

Frequency

Programs should develop benchmarks and periodically evaluate for continued consistency.

Quality Assurance

- Write precise procedures.
- Develop decision-making flow charts.
- Train staff with respect to addressing any deficiencies noted.
- Develop standardized data definitions for each data element. When applicable, develop a list of acceptable responses for a data element. Use drop-down windows to facilitate selecting from a list.
- Use technology to increase the accuracy of abstracting and data entry.

Tips

Keep a log of decision-making items and make sure it can be referred to easily. This is important for abstracting and coding procedures. Update procedure manuals, date-stamp all changes. When disease reporting rules or procedures change, make the changes effective as of the beginning of a calendar year.

Improving Quality through Timeliness Measurements

Purpose

To evaluate rapidity and readiness.

Background

All areas of case ascertainment and data collection affect how responsive the program is in meeting goals and objectives with respect to timeliness.

Method

The time interval between two or more points of interest is measured. Often the measurement is from one task to the next or from start to finish.

Outcome Measurements

Timeliness measurements can be used to evaluate and improve many areas within a surveillance program including productivity and program performance. Examples include:

- Reporting time lags. A measurement of the time it takes for a case report to be received-in or identified-to the birth defects program.
- *Case-finding process*. An evaluation of the time it takes to identify a case, review the medical record(s), abstract information, and complete the abstract.
- *Data processing time lags*. A measurement of how quickly information is processed for use.

Frequency

Timeliness can be evaluated readily. Tracking measurements can be monitored using software technology and developing date-posting fields. Queries, internal logs, and reports can facilitate this quality improvement method.

Quality Assurance

- Implement changes to case ascertainment procedures or processes to improve timeliness.
- Use laptops to reduce redundant steps.
- Work with data sources to improve consistency in reporting, including using electronic case reporting and Internet reporting.
- Develop computer transaction logs.

Tips

Evaluate the program's desired outcome measurements in relation to how long it takes to achieve them. Use the criteria in TACOMA, especially as they relate to improving timeliness. For example, the criteria 'oriented' and 'applicability' focus on selecting data variables that are important to the program. Include an evaluation of the reasons for unfinished case abstracts or case records. Data variables that consume a great deal of resources to collect should be re-evaluated for intent and usefulness.

Improving Quality through Data Source Evaluation

Purpose

To ensure that birth defect case reporting is complete, accurate, appropriate, and within the guidelines for timely reporting.

Background

Birth defects are found-at or reported-from data sources. Data sources vary in purpose, organizational structure, and scope.

Method

The source of the diagnostic information is evaluated for accuracy, completeness, and timeliness. This method may combine the methodology of other procedures, such as validity audits and timeliness measurements, and may also include re-case finding.

- *Accuracy*. The medical record, or other medical information report, is reviewed at the site or data source that reported the diagnosis or provided the diagnosis in a listing (see Validity Audits).
- Accuracy. For a large data source, such as hospital discharge data (an administrative data base), the audit may be designed to focus on a suspected hospital or unusual patterns of disease-code use.
- *Timeliness*. Time lags for reporting are evaluated per data source.
- Completeness. Passive case ascertainment utilizes the steps taken in active case-finding to identify all of the potential and confirmed cases of birth defects at the data source. This procedure is more difficult for passive case ascertainment to implement because of the staff resources needed to conduct comprehensive case-finding (see Chapter 6 on Case Ascertainment Methods).

Outcome Measurements

- *Completion of the descriptive assessment* of the data source.
- Refer to validity audits and timeliness audits.
- False positive rate. What is the level of diagnostic quality from a data source?
- *Completeness rate*. What is the rate of missed individuals with birth defects? These are individual cases that were not reported-to or identified-at the data source.
- Evaluation of data collection methods. Is the format used for reporting cases contributing to missed case reports?

Frequency

Each data source should be evaluated at least once to assess a level of quality.

Quality Assurance

- Use multiple data sources. One data source rarely provides comprehensive information.
- Use data sources that report a confirmed diagnosis.
- Involve the data source in discussions related to quality indicators. Develop mutually agreed-upon strategies for resolving issues.
- Encourage data sources to report cases in an electronic format, including using the Internet. This may improve timeliness and completeness. Confidentiality and privacy can be assured via encryption and other safeguards.

Improving Quality through Data Source Evaluation (continued)

Tips

Staff from programs using passive case ascertainment often review medical records in medical records departments, and some review autopsies at pathology departments. However, these staff usually do not engage in case-finding (i.e., combing through information sources to find potential cases of birth defects). Passive ascertainment staff should engage the data source in discussions prior to a case-finding audit. It is important to involve staff at the data source in planned activities to answer their questions. A contact person at the data source should be identified to ensure minimal disruption of normal work flow once the case-finding process begins.

Chapter 7 7-18 Data Quality

Improving Quality through Comparison/Verification Between Multiple Data Sources

Purpose

To compare diagnosis, and other information, that is reported-from or identified-at different data sources.

Background

Programs are encouraged to use multiple data sources for case ascertainment. A single data source is rarely able to provide comprehensive or accurate information

Method

- Compare information that is collected from multiple data sources in order to determine what information is accurate and complete. Examples include:
 - Confirm or invalidate a diagnosis based on a higher level of diagnostic expertise or clinical specialty. For passive case ascertainment this could mean that a diagnosis that is reported from a high-quality data source is considered to be confirmed or valid.
 - Clarify an incomplete or imprecise diagnosis. Conduct follow-up to gather better information.
 - Identify incomplete data fields on the case abstract or case record. Some data sources may not have complete information on a birth defect case, which results in an incomplete or deficient case report.
 - Update the case abstract or case record with more timely information.
 This includes address, names, and contact information.
- Develop procedures to identify duplicate case abstracts or case records in the database. Common situations that result in duplicate case abstracts or case records are mistakes with date of birth, use of multiple or incomplete names, and adoptions.

Outcome Measurements

- Rate the data sources. Assign 'quality' grades for specific criteria (e.g., diagnosis quality, complete address).
- Evaluate the value-added benefit that a data source provides. For example, if two data sources identify the same cases but one source provides a higher total volume of cases, evaluate the rationale for using both data sources.

Frequency

The use of multiple data sources is strongly encouraged. However, a surveillance program needs to understand the potential differences in quality among data sources and adjust procedures accordingly. In active case ascertainment, the comparison and verification of information can be done in an ongoing manner. In passive case ascertainment, where each case report may not be read by staff upon receipt, a benchmark should be established and key factors evaluated. At a minimum, comparison and verification should be done annually; otherwise the volume of inconsistencies or differences may turn into a resource-intensive effort to reconcile them. Computer technology greatly enhances a program's ability to systematically conduct comparison and verification procedures.

Improving Quality through Comparison/Verification Between Multiple Data Sources (continued)

Quality Assurance

- Combine or merge data that are collected or abstracted into a central case abstract or case record upon receipt. This minimizes the possibility of creating a duplicate abstract or record and reduces redundant staff work.
- Develop data linkage procedures for the large administrative, computerized data sets, such as vital records, hospital discharge data, hospital disease index, and Medicaid. Data linkage can also be developed to accommodate smaller clinic-based information systems, such as cytogenetics laboratories, genetic services, and specialty clinics. A key factor in data linkage is using standardized data variables (see Chapter 4 on Data Collection Variables).
- Develop decision-making and hierarchy models for use in comparison and verification of data elements. Programs should determine which data sources are considered a high-quality information source for specific data variables.

Tips

This QC method is enhanced by using computer technology and developing a systematic approach.

Chapter 7 7-20 Data Quality

Improving Quality through Computer Technology

Purpose To use technology in quality improvement efforts.

Background

Surveillance systems are information management systems whose operations are enhanced by computer technology (see Chapter 9 on Data Management and Security).

Method

- *Quality assurance*. Use software to prevent problems and enhance standardization.
 - O Build in range checks to prevent inaccurate abstracting and data entry. These checks can be created for any data variable with a defined parameter of acceptable or measurable values. Date range checks can be used for age, date of birth, date of fetal demise, date of death, LMP (date of last menstrual period). These become the dates that other dates (e.g., date of case report) are compared to for rationale. Other types of range checks are Apgar scores, gestational age, and birth weight.
 - Develop automated calculations and conversions for specific data fields.
 Examples include birth weight, time and LMP.
 - Promote the use of coded data. Develop codes for text information. This method can be applied to any data variable definition that has multiple acceptable responses. Examples include disease, geographic, race, and ethnicity codes. Programs can develop code sets for data sources, specific sites, types of procedures, family history, physicians, etc
 - *Use drop-down windows for data fields.* This approach is useful with long text entries and for text that has been converted to a code.
 - Use standard data collection variables (and data definitions), to accommodate record linkage and electronic transfers (see Chapter 4 on Data Collection Variables).
- *Quality control*. Develop procedures to detect, measure, and enhance effectiveness.
 - Perform logic edits. Review existing program documentation and syntax to ensure that the computer application is performing as intended. For example, when computer applications are used to convert or calculate data field values, make sure the results using the formula(e) are accurate.
 - *Create date-posting fields* to monitor timeliness.
 - Develop transaction logs. This is a method that tracks and dates additions, deletions, and other changes to the database.
 - *Create queries and reports* to track desired outcome measurements.
 - Develop methods, using key data variables, to find duplicate cases in the database.
 - Develop queries to identify problem situations. Examples include:
 - Some birth defects should not be counted due to prematurity or low birth weight.
 - Some ICD codes are problematic for birth defects.
 - Develop information management systems to improve the efficiency of program operations, including case ascertainment.
 - Develop methods to improve timeliness of case reporting. This includes using Internet reporting and other electronic methods, with appropriate security measures to protect confidentiality and privacy.

Improving Quality through Computer Technology (continued)

Outcome Measurements

Track measurements from the QC and QA methods that are developed.

Frequency

Once developed, computerized quality procedures can be run on a consistent and systematic timeframe. Systems and software also facilitate flexibility for ad hoc queries and reports. Information management systems are ongoing system enhancements.

Quality Assurance

Design, maintain, and update to:

- Prevent problems at the source
- Promote standardization
- Improve program efficiencies, including timeliness
- Facilitate data retrieval and analysis
- Assist in tracking measurements

Special considerations for passive case ascertainment programs

A dilemma that primarily affects passive case ascertainment programs is how to retain the integrity of the database, while also resolving data quality problems. In other words, how do programs identify and use accurate information, especially since the majority of diagnoses in the data base are accepted as reported (i.e., not confirmed by staff)? For example, if a diagnosis is reported from a data source and is determined to be inaccurate, or incomplete, it should not be counted in statistical analysis. However, for epidemiological and evaluation purposes, this diagnostic information (and the associated information that accompanies the case report) should not be deleted from the database. A method to resolve this issue could be to develop a mechanism (perhaps a data field) that identifies or flags a diagnosis that is not accurate (valid) or should not be counted. Programs are encourage to develop methods to resolve these kinds of issues in a way that best suits the program's needs.

Tips

- Before building a computerized data collection system evaluate the current manual data collection instrument to determine what works and what doesn't work.
- Avoid programming an on-line data collection system based on your hard copy instrument. Once programming is completed, it is often difficult to undo.
- Prior to developing and expanding the data base, evaluate the program's needs, i.e., how the data will be used, how the data will be accessible, data transfer, etc.

7.9 References

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Appendix 7.1 Data Sources Descriptive Assessment Tool

Appendix 7.1 Data Sources Descriptive Assessment Tool

Title of Data Source:

Evaluation Criteria Trait	Explanation and Description of Criteria	Specifics for This Data Source
Source or Site	The Source or Site is briefly defined	
Legal or Professional Mandates	Legal issues or professional requirements that govern or guide operations are described	
	Statutes or regulations Accreditation agencies (e.g.,	
	JCAHO)	
	 State licensing boards Federal agency requirements (e.g., medical participation agreements) 	
Mission or Objective	Purpose or reason that the Source or Site collects the information	
	How is the information used?	
Scope or Breadth	Time span or scope of time for the information collected	
	What time span does the Source or Site address? Point of time or follow-up capability?	
Operational Structure	Flow of information is described	
	Where is the information collected? Who collects the information?	
	 When is the information documented? 	
	How is the information documented?	
	 How is the information stored? 	

Appendix 7.1 Data Quality

Evaluation Criteria Trait	Explanation and Description of Criteria	Specifics for This Data Source
Type of Information Collected	Type of information collection is listed	
	Unique pieces of information that	
	the Source or Site collects are	
Acceptability and Datricyahility	listed Location of the information	
Accessibility and Retrievability	Location of the information	
	Accessibility to the information	
	Retrievability of the information	
	Data base, records merging, and other electronic applications capability	
	- Supularity	
Strengths as a Data Source/Site	The strengths and attributes of this Source/Site are described	
	Accuracy of the information	
	Completeness of the information	
	Timeliness of the information	
	Efforts and resources required	
	for case finding	
	Other qualities detailed	
Weaknesses as a Data Source/	The weaknesses and deficiencies of	
Site	the Source/Site are described	
	Accuracy of the information	
	Completeness of the information	
	Timeliness or time lags in case reporting	
	Effort and resources required to receive a case report	
	Other qualities detailed	
	Curor quantities detailed	
Liaisons and Partnerships	Key contact people/departments are identified	
	Ways to enhance cooperation	
	and partnerships are described	
Additional Comments	Issues to consider	
Additional comments	issues to consider	
References	Contact information	
	States with experience	
	Literature references	

Appendix 7.1 A7.1-2 Data Quality

Chapter 8 Statistical Methods

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8.1 Introduction

Statistics are useful to surveillance programs for:

- Summarizing and comparing surveillance data
- Assessing the potential role of chance or random variability
- ➤ Controlling for the effects of extraneous factors

The objective of this chapter is to present some common statistical concepts and tools that can be applied to surveillance data. For each tool, a definition is provided, along with background information, guidelines for use, how to calculate, and an example. This is a basic introduction only; more exhaustive treatment of these topics can be found in the reference literature.

In Section 8.2 of this chapter we discuss measures of birth defect occurrence. General issues relating to prevalence are discussed in Section 8.3, with the distinctions between crude prevalence, specific prevalence, and adjusted or standardized prevalence presented in Section 8.4. Various approaches to presenting and displaying descriptive epidemiology are described in Section 8.5, while confidence intervals and their calculation are discussed in Section 8.6. Finally, in Section 8.7 we discuss means to rule out straightforward explanations for observed changes in the prevalence of a birth defect. References cited in this chapter may be found in Section 8.8.

Chapter 8 8-1 Statistics

8.2 Measuring Birth Defect Occurrence

In carrying out basic epidemiologic and statistical assessment of birth defects occurrence, the analyst needs to decide what to count (issues of case definition are discussed in Chapter 3) and how to use those counts in calculations. This section presents some basic concepts, clarifies definitions with respect to analysis and reporting, and presents alternatives to the standard methods used to measure birth defects occurrence, birth prevalence.

8.2.1 Multiple Birth Defects in the Same Child

Analyses of birth defects surveillance data should be based on cases. An infant or fetus can have multiple birth defects and can be counted as a separate case for each defect. Thus, an infant/fetus with anencephaly and cleft lip should be counted as a case of anencephaly, and again as a case of cleft lip. When using this approach, it is important to recognize that the number of different cases cannot then be added to reach a total number of infants/fetuses.

When an infant/fetus has two or more conditions coded in the same category in an analysis, count it once only. For example, if an infant has atrial septal defect and ventricular septal defect. Count the infant once in tabulations for atrial septal defect, and once in tabulations for ventricular septal defect. Additionally, count the infant only once in tabulations for cardiac defects.

8.2.2 Counts, Ratios, Proportions, and Rates

The most common measures of birth defect occurrence are counts, ratios, proportions, and rates.

Counts. *Counts* present the simple enumeration of cases. Such information can be useful for health planning purposes, where it is important to measure the burden of birth defects on existing health care resources, to assess the need for additional resources, and for cluster investigations. However, simple counts of cases are not of value as a measure of disease risk, for which rates are necessary.

Ratios. A *ratio* is composed of one number (the numerator) divided by another (the denominator). Ratios can be useful for comparing the number of cases in one population group with the number in another. Proportions and rates (discussed below), and prevalence (discussed in Section 8.3) are special types of ratios.

FORMULA: A / B or numerator / denominator.

EXAMPLE: The sex ratio of cleft palate cases would be represented as notated below.

number of male cases with cleft palate number of female cases with cleft palate

Proportions. In a *proportion*, the cases in the numerator must be included in the denominator. A *percentage* is a proportion multiplied by 100. Proportions are useful for describing basic characteristics of surveillance program data. This can help with quality control.

FORMULA for a proportion: A/(A+B)

FORMULA for a percentage: $A \times 100 / (A+B)$

EXAMPLES

The *proportion* of abstracted records with errors would be:

the number of records with errors the total number of records

The *percentage* of abstracted records with errors would be:

the number of records with errors x 100 the total number of records abstracted

Rates. In epidemiology, *rates* express the frequency with which an event occurs (e.g., the number of new cases of disease) in a defined population in a specified period of time (Last, 1995).

FORMULA for incidence rate:

the number of new cases of a disease during a period of time population at risk

As will be discussed further in Section 8.3, although some investigators and studies report 'incidence rates' when talking about birth defects occurrence, there is general consensus that the information to determine incidence is not available (Sever, 2004). Therefore 'prevalence' or 'prevalence at birth' is the more appropriate terminology.

8.3 Calculating Prevalence at Birth

Prevalence expresses the number of existing cases of disease at a point in time divided by the total population. Prevalence is useful since it allows comparison between populations of different sizes. Prevalence may be measured at any time (e.g., X cases of spina bifida of any age on June 1 2003 divided by the entire population). However, for measuring occurrence of birth defects, it is most common to use prevalence at birth or birth prevalence. That is true even though many of the cases included may not have been live births.

Ideally, incidence rates would be used instead of prevalence to measure birth defect occurrence. *Incidence rates* measure the occurrence of new events that occur in a population, so the formula for incidence of a birth defect would be:

the number of new cases of birth defect A in an area and time period x the number of conceptions at risk of developing defect A in that area and time period

multiplier

10.000

Since the number of conceptions is unknown, as is the number of cases "lost" through spontaneous abortions, technically speaking we cannot determine incidence. Because of this, as noted above, most epidemiologists working in the area of birth defects use the term 'prevalence' to refer to birth defect occurrence. For a more complete discussion of this issue see Sever (2004).

8.3.1 Basic Calculation for Prevalence at Birth

Surveillance programs should measure birth defect occurrence using the following formula for birth prevalence. Note that both the numerator (number of cases) and the denominator (number of live births) always come from the same area and time period, that is, the same population. Usually the denominator is the number of live births to residents in the same geopolitical area from which the cases came during the same time period.

FORMULA for birth prevalence (expressed as cases of defect A per 10,000 live births):

the number of cases with birth defect A in an area and time period x the number of live births in that area and time period

EXAMPLE from Missouri:

193 cases with Tetralogy of Fallot statewide delivered in 1989-1995 x 10,000 532.592 live births

= 3.62 cases per 10,000 live births

Chapter 8 8-4 Statistics

8.3.2 Numerator and Denominator Issues

Counting cases – the numerator. For information on how to count cases for the numerator, see Section 8.2.1 on measuring birth defect occurrence (cases versus infants/fetuses).

Pregnancy outcomes included. Most often in analyses of birth defects surveillance data, the cases in the numerator are derived from all pregnancy outcomes collected by the program. These *may* include those listed below (see Chapter 3 on Case Definition).

- ➤ Live births
- > Spontaneous fetal deaths greater than or equal to 20 weeks gestational age (GA)
- > Spontaneous fetal deaths less than 20 weeks GA
- ➤ Induced terminations greater than or equal to 20 weeks GA
- ➤ Induced terminations less than 20 weeks GA
- Fetal deaths, unknown if spontaneous or induced, and/or of unknown gestational age

Sometimes analyses are restricted to certain pregnancy outcomes in comparing data from two surveillance programs that collect different pregnancy outcomes. For example, in the annual reports of EUROCAT (2002) and the International Clearinghouse for Birth Defects Monitoring Systems (2002) data are presented by live births plus stillbirths (late fetal deaths) and induced abortions (terminations of pregnancy), such that it is possible to make comparisons between prevalence based on comparable methods of case ascertainment (Sever, 2004).

The reports generated by the program should document which pregnancy outcomes are included in the numerator.

For the denominator, surveillance programs should use the total number of live births in the same area and time period from which the cases were ascertained. Although including induced and spontaneous fetal deaths would more closely approximate incidence rates calculated in other areas of epidemiology, it is impractical since these other pregnancy outcomes are often inaccurately counted compared to live births. In addition, these counts are small in comparison to the number of live births and are unlikely to affect prevalence to a large degree.

All the cases in the numerator (e.g., spontaneous and induced fetal deaths) may not come from the denominator (live births). For this and other technical reasons, birth prevalence is really a ratio and not a rate, although it is commonly referred to as a 'rate'.

Multiplier. The multiplier 10,000 is used for convenience, so that prevalence of most defects will have at least one digit to the left of the decimal place. Prevalence is expressed as 'X cases per 10,000 live births'. The most common multipliers for birth defects are 10,000 and 1,000.

EXAMPLE. 6.3 cases per 10,000 live births is easier for the reader to understand than 0.00063 cases (per birth).

Chapter 8 8-5 Statistics

8.4 General Types of Prevalence

As with rates and other measures of morbidity and mortality, there are three general types of prevalence. Prevalence can be categorized based on whether it:

- ➤ Applies to the whole population *crude prevalence*
- ➤ Applies to subgroups within the population *specific prevalence*
- Applies to the whole population, but adjusts for differing distribution of subgroups within the population *standardized or adjusted prevalence*.

Below we discuss each of these types of prevalence in turn.

8.4.1 Crude Prevalence

Definition Prevalence calculated for the entire population without regard to possible

subgroups within the population.

When to Use When a single, easily calculated number summarizing the occurrence of disease

in a population is desired.

How to Use For birth defects, the basic calculation above is applied to the entire population.

The area is usually the area covered by your birth defects surveillance program.

The number of cases with birth defect A in an area and time period x the number of live births in that area and time period

10,000

EXAMPLE

Birth Prevalence of Down Syndrome, Texas, 1996/97 Deliveries

Cases	Live Births	Prevalence*
355	300,431	11.82

^{*} cases per 10,000 live births Source: Ethen and Case, 2000.

8.4.2 Specific Prevalence

Definition Prevalence calculated for subgroups or *strata* within the population, such as age

groups, sex groups, or racial/ethnic groups. These then would be referred to as age-specific prevalence, sex-specific prevalence, or race/ethnicity-specific prevalence. The term 'stratified' is also used to refer to the prevalence among

such subgroups.

When to Use Specific prevalence is used in looking at disease occurrence in subgroups of a population. It is also used when sufficient data are available to define and

categorize the population of interest. In using specific prevalence, it is important

Chapter 8 8-6 Statistics

to consider how missing values (e.g., unknown maternal ages) would affect the interpretation of the prevalence data presented.

How to Use

For birth defects, apply the basic prevalence calculation above to each group of interest within the population. It is necessary to have the numerator and denominator from the same group of interest. The most common groups of interest for routine birth defects reports are based on:

- Maternal age at delivery
- Maternal racial/ethnic group
- Infant sex

However, specific prevalence can be calculated for any group for which numerator and denominator data are available.

Each grouping of a variable of interest is also called a 'stratum'. For example, common strata for maternal age at delivery are:

- Less than 20 years old
- 20 24 years old
- 25 29 years old
- 30 34 years old
- Greater than or equal to 35 years old

Calculating maternal age-specific prevalence would then yield five values.

It is helpful to define the groups or strata in the same way vital statistics are routinely reported for the population of the area. For example, live births in Texas are commonly reported for four maternal racial/ethnic groups:

- White (non-Hispanic)
- Black
- Hispanic
- Other

Thus, those categories are used for reporting race/ethnicity-specific prevalence values.

The FORMULA for calculation is:

the number of cases with birth defect A in group X in an area and time period x 10,000 the number of live births in group X in that area and time period

EXAMPLE

Maternal Age (years)	# Cases	# Live Births	Prevalence*
< 20	39	48401	8.06
20 – 24	53	83398	6.36
25 – 29	45	81442	5.53
30 – 34	84	57562	14.59
35 +	134	29574	45.31

^{*} cases per 10,000 live births Source: Ethen and Case, 2000.

8.4.3 Adjusted or Standardized Prevalence

Definition

Prevalence calculated for the entire population (the target population) that adjusts for (eliminates the influence of) possible differences in makeup between it and some standard or reference population. It is a summary measure that is a weighted average of the stratum-specific prevalence values.

There are two types of adjusted or standardized prevalence: direct and indirect. *Direct adjustment* uses specific prevalence derived directly from the target population (hence the name) and combines them using the age distribution of the standard population. The prevalence is generated that the target population would have experienced had it had the same age structure as the standard population. *Indirect adjustment* uses age-specific prevalence figures derived from the standard population but applies those to the age distribution of the target population. This technique produces the number of cases the target population would have experienced had it had the same age-specific prevalence as the standard population. The final result is usually expressed as a ratio of the cases observed in the target population divided by the number of cases expected based on this calculation.

When to Use

Adjustment is used to develop a single number summarizing the occurrence of birth defects within a population compared with some other population, removing the effect of differences between populations in the distribution of the factor adjusted for. An example would be to examine the occurrence of Down syndrome in a community near a hazardous waste site where the community has a larger proportion of older mothers than a comparison community or the state as a whole. The most common characteristics adjusted for in birth defects analyses are maternal age and maternal racial/ethnic group.

Use direct adjustment:

- When information is available on both the number of cases and the number of live births in each group/stratum/level of the factor being adjusted for (e.g., in each maternal age group); or
- To compare two or more target populations with each other (e.g., prevalence for an encephaly in 20 counties [20 target populations], standardized for maternal race-ethnicity group).

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Note that to compare two or more target populations, they must be standardized using the same standard population.

Use indirect adjustment when:

- Information is not available on the number of cases in each stratum of the factor being adjusted for;
- The comparison is between a target population and a standard and not with another target population (e.g., is the prevalence of anencephaly significantly different in County X compared to the whole state?);
- Statistical precision is very important (since stratum-specific prevalence used for direct standardization can sometimes vary widely if based on few cases); or
- The results are to be presented as an observed-to-expected ratio (although a prevalence can be calculated).

The following instructions are based on adjusting for maternal age groups. Each age group is called a 'stratum'. The same process would be used when adjusting for other characteristics, for example, race or ethnicity.

Direct adjustment. For direct adjustment follow the steps below.

- 1. Decide on age-group categories (strata) that can be applied to both the target and standard populations.
- 2. Calculate age-specific prevalence for each stratum of the *target* population. Do not use the multiplier (10,000 or 1,000) for this calculation. However, the multiplier may be used for presenting the age-specific prevalence values.
- 3. Multiply each prevalence by the number of live births in the same stratum of the *standard* population. This gives the number of cases expected in each stratum of the standard population, had it experienced the same age-specific prevalence as the target.
- 4. Add up the number of expected cases across all strata.
- 5. Divide the total number of expected cases by the total number of live births in the *standard* population and multiply by your multiplier (e.g., 10,000).

This is the 'directly standardized birth prevalence'.

Indirect adjustment. For indirect adjustment follow the steps below.

- 1. Decide on age-group categories (strata) that can be applied to both the target and the standard populations.
- 2. Calculate age-specific prevalence for each stratum of the *standard* population. Do not use the multiplier (10,000 or 1,000) for this calculation. However, the multiplier may be used for presenting the age-specific prevalence.
- 3. Multiply each prevalence by the number of live births in the same stratum of the *target* population. This gives the number of cases expected in each stratum of the target population, had it experienced the same age-specific

How to Use

prevalence as the standard.

- 4. Add up the number of expected cases across all strata of the *target* population.
- 5. Divide the total number of observed cases in the *target* population by the calculated total number of expected cases.

This is the 'standardized birth prevalence ratio', sometimes called the 'standardized observed-to-expected ratio'. When applied to mortality, the result is called the 'standardized mortality ratio' or SMR.

EXAMPLES. The Texas Birth Defects Monitoring Division dealt with a cluster of Down syndrome in a three-county area in Texas among deliveries in 1992–1994. Down syndrome is influenced strongly by maternal age. Thus it was necessary to adjust for maternal age to see whether the excess was still apparent when possible differences in maternal ages between these three counties and the state of Texas were removed. The three counties (1992–1994 deliveries) make up the *target population*, and the entire Texas Birth Defects Registry area (1996–1997 deliveries) the *standard population*. The years 1996–1997 were used because those were the first years with data published for most of the state.

Direct adjustment. The crude Down syndrome prevalence for the three counties during 1992–1994 was 31.97 cases per 10,000 live births. **Steps 1-4** are presented in the following table.

Maternal		Target		Stand	dard
Age (years)	# Cases	# Live Births	Prevalence*	# Live Births	Expected # Cases
	(A)	(B)	(A) / (B) = (C)	(D)	(C) x (D)
< 20	3	1032	0.00290698	48401	140.701
20 – 24	3	1666	0.00180072	83398	150.176
25 – 29	3	1498	0.00200267	81442	163.101
30 – 34	4	1028	0.00389105	57562	223.977
35 +	5	407	0.01228501	29574	363.317
Total	18	5631	0.00319659	300377	1041.272

^{*} Expressed as cases per live birth. To express it the usual way, multiply by 10,000.

Step 5

total number of expected cases in standard population x 10,000 total number of live births in standard population

= 1041.272 x 10,000 300377

= the standardized prevalence

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Indirect adjustment. **Steps 1-4** are presented in the following table. There are known to be 18 cases of Down syndrome in the target population, but the ages of their mothers may not be known.

	TARGET		STANDARD				
Maternal Age	# Live Births	# Cases	# Live Births	Prevalence *	Expected # Cases		
(years)	(E)	(F)	(G)	(F) / (G) = (H)	(E) x (H)		
< 20	1032	39	48401	0.00080577	0.83155307		
20 – 24	1666	53	83398	0.00063551	1.05875441		
25 – 29	1498	45	81442	0.00055254	0.82770561		
30 – 34	1028	84	57562	0.00145930	1.50015635		
35 +	407	134	29574	0.00453101	1.84411983		
Total	5631	355	300377	0.00118185	6.06228927		

^{*} Expressed as cases per live birth. To express it the usual way, multiply by 10,000.

Step 5

 $\frac{total\ number\ of\ observed\ cases\ in\ target\ population}{total\ number\ of\ expected\ cases\ in\ target\ population}\ x$

$$= \frac{18}{6.0623} \quad x \quad 10,000$$

= 2.97

= standardized birth prevalence ratio or standardized observed-to-expected ratio

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8.5 Descriptive Epidemiology

Background

Surveillance data allow the description of the occurrence of birth defects in terms of the basic epidemiologic parameters of time, place, and person. In doing so, comparisons can be made among these different parameters (e.g., comparing the prevalence of anencephaly among different maternal ages).

Note that descriptive epidemiology is really just the presentation of specific prevalence where the strata are time periods (e.g., year of delivery), areas (e.g., counties), or personal characteristics (e.g., maternal age groups or maternal racial/ethnic groups). See Section 8.4.2 above on calculation of specific prevalence.

When to Use

To describe patterns of birth defect occurrence.

How to Use

Choose the most appropriate measure of birth defect occurrence. This will usually be prevalence, but in some circumstances it may be counts. For example, in planning for services, the actual number of children in a population born with a defect, such as cleft lip, may be more important than the prevalence. Analyze the chosen measure according to the basic epidemiologic parameters of time, place, and person (Teutsch and Churchill, 2000; Seiffert, 1994). Tables, graphs, and maps are very useful in presenting data in an understandable form; suggested approaches can be found in Teutsch and Churchill (2000).

Time. Surveillance programs should clearly state the relevant time period of study from which cases are drawn. This is usually based on 'date of delivery'. Other options include 'estimated date of conception', 'estimated date of delivery', 'date of diagnosis', or 'date of incorporation into the surveillance database'. Common analyses of birth defect occurrence by time include:

- Prevalence by year: to look for long-term trends
- Prevalence by season or month within the year: to look for seasonal patterns
- Counts of cases by date of delivery or estimated date of conception: to look at birth defect clusters

Place. Reports should specify the geographic area of coverage for the prevalence presented in the report. Common analyses of birth defect occurrence by place include:

- Prevalence by state, county, or region
- Prevalence by zip code or census tract
- Spot maps of cases showing residence at delivery or conception

Person. Common analyses of birth defect occurrence by person include:

• Prevalence by maternal age (age of the mother at the time of delivery)

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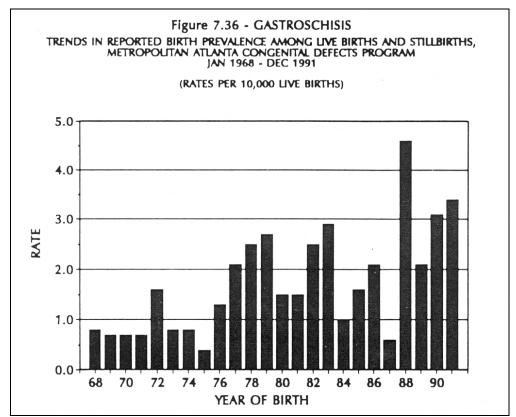
- Prevalence by maternal race/ethnicity (the most common definition is as stated by the mother)
- Prevalence by infant/fetus sex.

EXAMPLES

Time trend in the prevalence of gastroschisis over several years. The numerator (number of cases) and denominator (number of live births) for each year is determined. The prevalence for each year is calculated as:

the number of cases with gastroschisis in Metro Atlanta in year X the number of live births in Metro Atlanta in year X

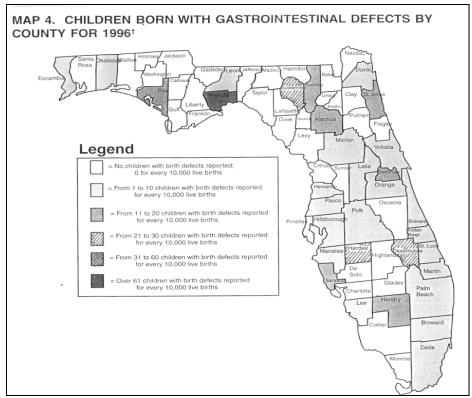
10,000



Source: Centers for Disease Control and Prevention, 1993, as cited in James et al., 1993.

Occurrence of birth defects by *place*, specifically, county prevalence of gastrointestinal defects. Numerators and denominators were collected for each county in Florida for deliveries in 1996. The results are presented in a chloropleth map (a method of mapping to display quantitative information where the areas [e.g., counties] are colored or shaded according to the value of some variable [e.g., gastroschisis prevalence]).

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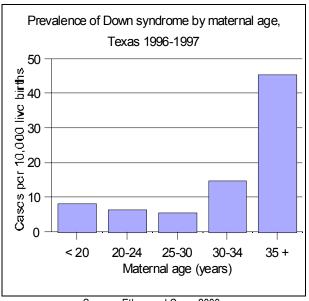
Source: Florida Department of Health, 1999.

Occurrence of birth defects by *person*. Numerators and denominators were determined for five maternal age groups. Stratum-specific prevalence is calculated and presented in a table and vertical bar graph.

Prevalence of Down syndrome by maternal age,

Maternal Age (years)	Cases	Live Births	Prevalence*
< 20	39	48,401	8.06
20-24	53	83,398	6.36
25-30	45	81,442	5.53
30-34	84	57,562	14.59
35 +	134	29,574	45.31

*cases per 10,000 live births Source: Ethen and Case, 2000.



Source: Ethen and Case, 2000.

8.6 Confidence Intervals

In this section, we first discuss confidence intervals generally (Section 8.6.1). We then discuss the use of confidence intervals in comparing prevalence values (Section 8.6.2).

8.6.1 About Confidence Intervals

Definition

An interval around a statistic that contains the true underlying value of the statistic a certain amount of the time. For example, a 95% confidence interval for the prevalence of spina bifida will contain the true underlying value of the spina bifida prevalence 95% of the time.

The interval is bounded by an upper confidence limit and a lower confidence limit.

Background

The birth prevalence for a particular defect is estimated by the number of cases with the defect of interest ascertained from the population, divided by the total number of live births, and multiplied by some factor such as 10,000. This number is the best estimate of the true birth prevalence, which can never be known with certainty. To provide an idea about the precision of the estimated prevalence, a range of values is often calculated that is highly likely to contain the true prevalence. This range of likely values is called a *confidence interval*.

A confidence interval is calculated in such a way that, if the same procedure were to be repeated a large number of times, the proportion of intervals that contain the true prevalence would equal the confidence level. So, for example, if we choose a 95% confidence value, then 95% of all those confidence intervals will contain the true, but unknown, prevalence.

The 95% value is the conventionally used confidence interval. However, sometimes people choose other values, such as 90% confidence intervals (which are narrower than 95% confidence intervals) and 99% confidence intervals (which are wider).

Estimated confidence intervals for any given level (e.g., whether 90%, 95%, or 99%) will be narrower when their prevalence values are based on more cases.

Confidence intervals only measure random error, for example, when the occurrence of a birth defect fluctuates up and down from year to year by chance. They do not address systematic error or bias. Let's say, for example, one wanted to compare two surveillance programs. If program A does not ascertain cases from prenatal diagnosis clinics and thus consistently misses cases, while program B does ascertain cases from such clinics, a confidence interval around the prevalence of birth defects from program A should not be interpreted as accounting for the missing cases. The confidence interval merely addresses random fluctuation in the cases from program A.

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When to Use

Confidence intervals may be calculated for any summary statistic, e.g., for proportions. However, we will only discuss confidence intervals for birth prevalence since that will be the most common statistic presented by birth defects surveillance programs. To learn about calculating confidence intervals for proportions or other types of rates, please consult one of the statistics books listed in the reference section (e.g., Dawson and Trapp, 2001; Fleiss et al., 2003; Snedecor and Cochran, 1989).

Confidence intervals can be used whenever calculating birth defect prevalence, although their use is controversial. This issue is discussed extensively in a recent commentary (Costa and Kirby, 2003) and a theory and methods paper (Correa-Villasenor et al., 2003) on the use of confidence intervals and on errors and undercounting in birth defects surveillance data. The interested reader is referred to these publications.

Why to Use

From a theoretical viewpoint, prevalence (e.g., X cases per 10,000 among deliveries in 1999) can also be considered to be just one sample in time, and confidence intervals give an idea of the range of values within which the true value is likely to be found. From a practical viewpoint, confidence intervals are particularly useful when dealing with small numbers of cases or where the birth defect prevalence for one group will be compared with that of other groups. This is because confidence intervals can help minimize reader concern about prevalence values that appear high or different when this is most likely due to random fluctuation. Some states have found this to be particularly helpful, for example, when looking at prevalence for counties, areas smaller than counties, or racial/ethnic groups. While the best way to compare prevalence values between different areas is always to use a statistical test, it is not practical for a surveillance system to anticipate all the comparisons readers will want to make. Confidence intervals thus provide a quick way for readers to get a rough idea of the impact of chance on the data.

Why Not to Use

Some surveillance programs ascertain all cases of birth defects, so that the prevalence reported is not just a sample but is considered to reflect the underlying true prevalence. Therefore, the use of confidence intervals is considered by many to be irrelevant. Calculating confidence intervals also increases statistical work for program staff. Finally, some data users, for example community groups or the media, may find confidence intervals confusing.

How to Calculate

Upper and lower 95% confidence limits are shown in the table below; they are the end points of the corresponding confidence intervals. Note that calculation of confidence intervals for prevalence is merely the calculation of confidence intervals for the number of cases – the denominator portion of prevalence does not change.

For a prevalence based on a small number of cases. For small numbers of cases (arbitrarily defined here as fewer than 30), use the Poisson distribution since birth defects are considered to be rare events.

The easiest way to use the Poisson distribution is to refer to a table that provides the upper and lower 95% confidence limits for an observed number of *cases* (reproduced below for up to 29 cases). Then follow Steps 1 through 3.

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Table of 95% Confidence Limits for the Number of Cases, for 1-29 Cases, Based on the Poisson Distribution

Number of Cases	95% Confidence Limits			
Number of Cases	Lower	Upper		
0	0.0000	3.6889		
1	0.0253	5.5716		
2	0.2422	7.2247		
3	0.6187	8.7673		
4	1.0899	10.2416		
5	1.6235	11.6683		
6	2.2019	13.0595		
7	2.8144	14.4227		
8	3.4538	15.7632		
9	4.1154	17.0848		
10	4.7954	18.3904		
11	5.4912	19.6820		
12	6.2006	20.9616		
13	6.9220	22.2304		
14	7.6539	23.4896		
15	8.3954	24.7402		
16	9.1454	25.9830		
17	9.9031	27.2186		
18	10.6679	28.4478		
19	11.4392	29.6709		
20	12.2165	30.8884		
21	12.9993	32.1007		
22	13.7873	33.3083		
23	14.5800	34.5113		
24	15.3773	35.7101		
25	16.1787	36.9049		
26	16.9841	38.0960		
27	17.7932	39.2836		
28	18.6058	40.4678		
29	19.4218	41.6488		

Source: Diem and Lentner, 1970.

Step1. Calculate prevalence.

the number of cases with birth defect A in an area and time period $\, x \,$ the number of live births in that area and time period

10,000

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Step 2. Look up the lower 95% confidence limit for the number of cases with birth defect A. Using this new number in the numerator, calculate the lower 95% confidence limit for prevalence:

Step 3. Look up the upper 95% confidence limit for the number of cases with birth defect A. Using this new number in the numerator calculate the upper 95% confidence limit for prevalence.

EXAMPLES using data from the California Birth Defect Monitoring Program 1983-1986.

Birth Defect	Cases		Number of	Prevalence ⁶	
Dil til Delect	Number ¹	95% Cl ² Live Births		Value ⁴	95% CI⁵
Anophthalmia	18	10.67 - 28.45	452,287	0.40	0.24 - 0.63
Glaucoma	27	17.79 - 39.29	452,287	0.60	0.39 - 0.87

Notes

Source: Croen et al., 1990.

- (1) Number of cases ascertained from surveillance
- (2) 95% confidence interval for that number of cases
- (3) Number of live births derived from vital records
- (4) Prevalence = $[(1)/(3)] \times 10,000$
- (5) 95% confidence interval for the prevalence
- (6) Prevalence expressed as cases per 10,000 live births

For a prevalence based on a large number of cases. For a large number of cases (arbitrarily defined here as 30 cases or more), use the normal distribution. Why? As the number of cases grows larger, the Poisson distribution approximates (i.e., looks more and more like) the normal distribution. The formulae below are approximations for calculating confidence intervals using the normal distribution (Rothman and Boice, 1982, p. 29, formula 19).

Shorthand: let c = number of cases

b = number of live births

1. Calculate the lower confidence limit using the following:

Lower 95% CL for prevalence =
$$c \times \left(1 - \frac{1}{9c} - \frac{1.96}{3} \sqrt{\frac{1}{c}}\right)^3 / b \times 10000$$

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2. Calculate the upper confidence limit using the following:

Upper 95% CL for
$$\text{prevalence} = (c+1) \times \left(1 - \frac{1}{9(c+1)} + \frac{1.96}{3} \sqrt{\frac{1}{(c+1)}}\right)^3 / b \times 10000$$

- 3. To determine the 90% confidence limits, replace 1.96 with 1.645. To determine 99% limits, replace 1.96 with 2.575.
- 4. To obtain confidence limits for the number of cases instead of the prevalence, apply the formulae but do not divide by births (*b*) or multiply by 10,000.

EXAMPLES using data from the California Birth Defect Monitoring Program, 1983-1986.

Birth Defect	Cases		Number of	Pre	valnce ⁶
Birtii Delect	Number ¹	95% Cl ²	Live Births ³	Value ⁴	95% CI⁵
Aortic stenosis	73	57.22 - 91.79	452,287	1.61	1.27 - 2.03
Cleft palate	320	285.89 - 357.05	452,287	7.08	6.32 - 7.89

Notes

Source: Croen et al., 1990.

- (1) Number of cases ascertained from surveillance
- (2) 95% confidence interval for that number of cases
- (3) Number of live births derived from vital records
- (4) Prevalence = $[(1)/(3)] \times 10,000$
- (5) 95% confidence interval for the prevalence
- (6) Prevalence expressed as cases per 10,000 live births

Software support. For a few prevalence values, confidence limits (and hence the resulting intervals) can be calculated by hand or using statistical software such as PEPI. One program in PEPI (POISSON) gives the table values for any number of cases; for a large number of cases it gives a normal approximation. PEPI software and documentation are available at:

http://sagebrushpress.com//pepibook.html

For many prevalence values, it is useful to write programs, for example in SAS, to calculate the confidence limits along with the prevalence.

8.6.2 Comparing Prevalence Values Using Confidence Intervals

The best way to compare prevalence values (e.g., for different maternal ages) is to do a statistical test; one type of statistical test is described in the second section below ("When both prevalence values are based on 30 or more events"). However, in the first section below we present a quick method recommended by the National Center for Health Statistics (NCHS) that works better when one of the prevalence values is based on fewer than 30 cases. Note that the NCHS-recommended method is conservative (i.e., there will be fewer statistically significant differences than would be found by actually performing a statistical test).

When one of the prevalence values is based on fewer than 30 cases. First compute the 95% confidence intervals for both prevalence values. Check to see if those intervals overlap. If they *do* overlap, the difference is not statistically significant at the 95% level. If they *do not* overlap, the difference is indeed 'statistically significant' or unlikely to be explained by chance alone. (Method recommended by NCHS in Ventura et al. [2000].)

EXAMPLE

The prevalence of holoprosencephaly among African-American women in Texas (2.39 cases per 10,000 live births) is over three times higher than among White women in Texas (0.78 cases per 10,000 live births). Is the difference statistically significant? First compute the 95% confidence intervals.

Holoprosencephaly Among Two Race/Ethnic Groups in Texas, 1996/97

Group	Cases	Live Births	Prevalence*	95% CI for Prevalence
African American women	7	29,254	2.39	0.96 - 4.93
White women	8	102,193	0.78	0.34 - 1.54

^{*} cases per 10,000 live births

Source: Ethen and Case, 2000.

These two confidence intervals overlap. Thus, based on this approach, the difference between prevalence of holoprosencephaly in African-American women compared to White women is not statistically significant.

When both prevalence values are based on 30 or more events. This approach is based on calculating the confidence interval for the difference between the two prevalence values. If this interval includes 0.00, then the difference in the values is not considered to be statistically significant. Since this approach uses information from both prevalence values at the same time, it is more statistically powerful than the NCHS-recommended method. That is, if a difference truly exists, this approach will identify that more often than will the NCHS-recommended method. This approach uses the standard error for the difference between the two prevalence values (Rothman, 1986, p. 170, formulae 11-15).

Statistical software like the RATES2 program within the PEPI package can also be used to calculate this confidence interval.

Shorthand: let RD = higher prevalence - lower prevalence

 c_1 = number of cases used to calculate the first prevalence

 c_2 = number of cases used to calculate the second prevalence

 b_1 = number of live births used to calculate the first prevalence

 b_2 = number of live births used to calculate the second prevalence

1. Calculate the lower confidence limit using the following:

$$\text{Lower 95\% CL for prevalence difference} = RD - \left(1.96\sqrt{\frac{c_1}{b_1^2} + \frac{c_2}{b_2^2}} \times 10000\right)$$

2. Calculate the upper confidence limit using the following:

Upper 95% CL for prevalence difference =
$$RD + \left(1.96\sqrt{\frac{c_1}{b_1^2} + \frac{c_2}{b_2^2}} \times 10000\right)$$

- 3. To obtain 90% confidence limits, replace 1.96 with 1.645. To obtain 99% limits, replace 1.96 with 2.575.
- 4. If the confidence interval *does not include 0.00*, then the difference would occur by chance less than 5 times out of 100 (for 95% confidence intervals); i.e., the two prevalence values are significantly different.

EXAMPLE

Is there a statistically significant difference between African-American and Hispanic women in the prevalence of births with atrial septal defects?

Atrial Septal Defect Among Two Race/Ethnic Groups in Texas, 1996/97

Group	Cases	Live Births	Prevalence *
African-American women	133	29,254	45.46
Hispanic women	835	160,094	52.16

* cases per 10,000 live births

- Source: Ethen and Case, 2000.
- 1. The lower 95% confidence limit for the prevalence difference = -1.80.
- 2. The upper 95% confidence limit for the prevalence difference = 15.20.
- 3. The 95% confidence interval for the prevalence difference thus = -1.80 to 15.20.
- 4. The interval includes 0.00. Therefore, the difference between the two prevalence values is not statistically significant at the 95% confidence level; i.e., there is no statistically significant difference.

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8.7 Issues to Consider If Data Reveal Unusual Patterns

It is very important to rule out relatively straightforward explanations for a change in the birth prevalence of a birth defect. Among the more common reasons are:

- Changes in medical diagnoses and technologies
- ➤ Changes in reporting that lead to changes in case ascertainment
- ➤ Changes in the population at risk (focus on age, period, and cohort effects)
- > Random variation

Please refer to Kallen (1998, pp. 83-87) for a more extended discussion of the issues identified above.

The analytical capabilities of the surveillance system should support evaluation of the likelihood of these factors being responsible for observed changes in prevalence. Although the remainder of this section addresses "changes" in birth defect prevalence over time, it can also be applied to "differences" in birth defect prevalence between areas.

8.7.1 Changes in Medical Diagnoses and Technologies

To detect changes in medical diagnoses, it is important to compare isolated and multiple birth defects cases. Minor changes in the way a condition is diagnosed or reported can affect the coding and classification of specific birth defects. For example, the prevalence of neural tube defects among live-born infants may have declined during the 1980s from levels reported in the 1970s, due to the development and widespread availability of prenatal diagnostic tests, such as maternal serum alpha-fetoprotein screening and ultrasonography. The severity of spina bifida cases may be less today than in the 1970s due to the selective therapeutic termination of the more severe cases, which are more likely to be identified prenatally.

The birth prevalence of some disorders may increase due to new technologies. For example, fragile X syndrome, a chromosomal breakage disorder, is diagnosed much more often today than 10 years ago and was unknown 20 years ago.

8.7.2 Changes in Reporting and Case Ascertainment

Re-verification that the frequency is an un-duplicated count is also appropriate. Many suspected "clusters" reported by the media or concerned citizens involve multiple counting of the same cases. With a birth defects surveillance program that ascertains cases from multiple sources, it is important to ensure that each case is counted only once, even if reports are received from several health care providers or delivery settings. The same is true of changes in reporting. As hospitals shift to computerized diagnostic indices, reporting artifacts could decrease the numbers of cases of specific birth defects while increasing others. This is because a limited number of ICD-9-CM codes are retained in the index, and conditions that appear to be minor in the eyes of the medical records clerk may be omitted. If surveillance staff rely exclusively on the diagnostic indices to identify charts to abstract, some conditions may be missed. See also Chapter 5 on Classification and Coding and Chapter 6 on Case Ascertainment Methods.

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8.7.3 Changes in the Population at Risk

Population characteristics can be controlled for by using stratum-specific prevalence, age-adjustment, and similar methods. However, as most statistical surveillance methods are based on the frequency of events rather than on proportions, rates, or prevalence, separate analyses will need to be conducted to rule out changes in the population at risk. Analysts who routinely examine birth defects surveillance data will have access to detailed, current population estimates and should examine the demographic and reproductive health characteristics of all women giving birth to identify changes in the population at risk.

8.7.4 Random Variation

It is also possible, and in fact very likely, that an observed difference in the frequency of a specific birth defect is due to random variability. With relatively low birth prevalence, cases of a particular condition will be quite rare, and the coincidence of two or more cases in space and/or in time may be just that: a coincidence. Confidence intervals are one way to address random variation.

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Chapter 9 Data Management and Security

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9.1 Introduction

This chapter is designed to provide basic guidance on the mechanical and administrative aspects of establishing and operating a birth defects surveillance program, covering a range of topics related to the development of an efficient, effective, and secure program. This chapter is intended to serve as a guide to planning the development of a new surveillance program, as well as to the review of practices and procedures in place within existing programs.

Issues covered in this chapter include computer hardware and software, data capture procedures, transmission of data, data file management, personnel management, physical aspects of the surveillance program office and, finally, data confidentiality and security considerations. We discuss the functionality of a data processing system in Section 9.2, followed by more detailed discussions of hardware and software in Sections 9.3 and 9.4, respectively. Data management is introduced in Section 9.5 on process standards, while the specific topics of data entry (Section 9.6), record linkage (Section 9.7), and record consolidation (Section 9.8) are described in further detail in subsequent sections. The importance of ongoing communication with data sources to identify and correct emerging problems is discussed in Section 9.9, and in Section 9.10 we address physical security and confidentiality issues.

9.2 Functional Data Processing System Features

The functionality of the system for processing and managing surveillance data must be able to support all the necessary processes and activities required by a program. This chapter is intended to develop the key considerations and capabilities that are applicable to surveillance operations. There is a wide variation in case volume, approach to data collection, budgets, and goals of various birth defects surveillance programs. Overall mission, size, and scope will determine the best combination of procedures and features for a given program.

The basic operations of a birth defects surveillance system can be accomplished using minimal computer hardware, software, and systems. The characteristics outlined below provide a broad scope of useful features and capabilities.

Computerized data collection. As case reports on birth defects cases are received, data should be captured within an electronic database designed to maximize a program's ability to manage the surveillance system and utilize the resulting data.

A program's capacity to receive computerized data from reporting facilities and other sources can ease the burden of case reporting and reduce or eliminate the need for recapturing already automated data. Improving the efficiency of data collection can minimize effort in the reporting facility and at the surveillance program, while reducing errors often due to "re-automation." Increased efficiency can also improve relations with reporting facilities and support compliance with reporting requirements. Below we present critical considerations related to accepting and processing electronic reports from reporting facilities.

- > Reporting case data within electronic files, rather than paper reports, requires the exchange of detailed information on data submission requirements and on the characteristics of the files provided. Design issues, such as file formats and structures, and coding schemes must be understood to ensure accurate data exchange.
- Any limitations in the reporting facility's computer database must be identified to ensure that submitted data can meet programmatic needs. Any shortcomings or incompatibilities between the facility's system and reporting requirements must be recognized and addressed. Examples of such concerns include facility data systems that are missing standard (required) data items or that code a given item using coding rules that are not entirely compatible with the program's coding schemes.
- As the data systems used to generate the data files are revised, any effects that such changes may have on the submitted data should be identified. Information of this type must be communicated by the reporting facility to the surveillance system.
- > Submitted data must be reviewed for quality control. This review should compare the data submitted with source documents or files to validate that the data are being represented faithfully within the surveillance database. The review should identify any data distortions caused by differences in processing systems, coding structures or rules, and conversion routines used to build the extract file or to import the data into the surveillance program.

Transmission of electronic data and data telecommunication. Below we present considerations related to transmission of electronic data.

- ➤ If case reports are accepted as electronic files, a standard format, file structure, code structure and medium for submission, i.e., tape or disk, must be developed and documented.
- ➤ Programs receiving passive case reports may elect to accept data in formats and code structures that follow the reporting facility's database structure and rules. In this case the facility whether a hospital, a diagnostic laboratory, or other facility must be expected to refer to the standard for submissions and provide the data in a format and code structure that is compatible with and convertible into the standard format for reporting cases to the surveillance system.
- ➤ Whether converted or nonstandard data files are supplied, each facility must identify any compatibility/consistency problems apparent between the source system and the standard.
- > Secure methods for delivery of forms and data files need to be recommended by the surveillance program and followed by each facility.

Various modes for data entry. Below we present considerations related to data entry modes.

- To automate the information received in the form of paper case abstracts, a data capture mechanism is required. Approaches to accomplish this task include:
 - Classic data entry by keying data into a fixed format file
 - Optical scanning
 - Data capture through the use of custom screens
- > Principles associated with the data entry process, which will ultimately enhance efficiency and data quality, include:
 - Standardizing data review, query, and preparation procedures
 - Verifying the keyed data
 - Editing at the point of entry
 - Editing of the completed input file
- In addition to capturing case data internally, consideration should be given to developing software that enable facilities to report case data electronically. Providing such software to the reporting facilities enables them to use standard file formats and coding and editing procedures for the data they submit. Editing the data at the point of entry, in particular, can reduce the need for later follow-back.

Receipt and integration of new case data while retaining data on reporting activities. Below we present considerations related to the integration of new case data.

- > To the degree practical, the program needs to be able to receive submitted data for processing in the form of electronic files, paper submissions, and, potentially, through web-based or other direct data entry across secure connections.
- Integration of data into the system should be done such that integrity of the individual reports received is maintained. This allows the surveillance system to document the data source properly and to monitor reporting quality.

Ability to link surveillance data to new reports and files from other data sources. Below we present considerations related to linkage of surveillance data to new reports and files from other data sources.

- The program must have appropriate software and system computing capacity to screen incoming data against existing surveillance data in order to identify accurately duplications in reporting.
- The program must have the ability to implement a variety of strategies to link surveillance data with data from other sources in a manner that allows cases within other data sets to be identified as important. Through this means the program can augment the surveillance data base with information on cases identifiable through other data systems, as well as acquiring new case reports. This capability is essential if the surveillance program is to facilitate research studies.

Ability to modify system easily and inexpensively. The system used must provide flexibility with respect to systems modifications, edit specifications, and other data handling processes, as well as permitting modifications of code structure and data set variables within the database. When possible, programs should avoid developing the database using software systems that require considerable time and expertise to modify. It is best if modifications are controlled and can be made by birth defects surveillance staff.

Ability to handle updates. The system must provide an easy way to update registry data as new information on cases is received. Updates may include information on additional hospitalizations, further diagnostic work, or corrections to earlier reports.

Editing data. The system must support data editing at various steps throughout the data collection process. Data editing should be carried out as data are collected, processed, and incorporated into the program's database. Conducting edit checks as early as practical in the data collection process is an efficient way of improving data quality. Key stages for data editing include:

- > At the point of data abstraction
- During data entry
- > As files of new report data are prepared
- ➤ As case files are updated with new cases
- > As additional data on known cases are added

Common edit procedures include field code range checks, table look-up of diagnostic and other codes, inter-field consistency checks, and editing across records for individual cases.

Report preparation. Staff must have the computer capability and training to conduct statistical analyses; to interpret the resulting statistical information; and to prepare text, tables, and graphics in the form of reports. Examples of statistical analyses include establishing basic case counts and rates, developing summary data on treatment information, reviewing prevalence trends, adjusting rates, calculating variance components and standard errors, and developing measures of the observed and expected prevalence of specific conditions.

On-line case queries. The database should be readily accessible to staff using different types of information to identify specific cases.

Easy maintenance of reference tables/files. The various reference files used to process and edit incoming reports and to develop statistical data on those reports must be easy to maintain and update. Such reference files may include tables of diagnostic and procedures codes, code groupings, geographic code dictionaries, hospital and laboratory code dictionaries, among others.

Extracting files/subsets. The capacity should exist to generate readily subset files of the surveillance data. This capacity should allow inclusion of data on cases selected using a variety of criteria and inclusion of specific variables for selected cases. File subsets often are needed for statistical analyses, quality control work, field site visits, and other uses.

Quality control information on data sources, amounts, and quality. To monitor case reporting timeliness and quality, the system will need to store sufficient information to support calculations of reporting timeliness and other data reporting quality measures. The system must allow for assessment of reporting quality overall and by reporting source.

Systems security, administration, and backup. The system must include features to protect data and programs from loss due to systems failure or user error and to maintain the confidentiality of patient and provider data. The computer system must provide a secure environment with security features designed and enabled to protect data from inappropriate access. Such measures must include a system of user name and passwords, along with a system to control the access of users to the computer server and drive locations where data are stored. These must be updated promptly with staff changes.

Program staff must be able to control or oversee these system administration activities. The system must also provide redundant back-up procedures to protect against system failure. This should include back-up and recovery procedures with regular and reliable copying of existing surveillance data and systems to tape or disk.

Archiving of data and systems. The surveillance data and the systems used to develop and maintain the data must be archived according to a predetermined schedule to protect against catastrophic loss. Archiving procedures must ensure appropriate preservation of submitted case abstracts and the routines used to process abstracts and to analyze case information. Archiving should also encompass statistical analyses, special studies, and the procedures used in those studies.

Cost effectiveness. The computer system and hardware used must be selected to fit both the needs and the budget of the surveillance program. The initial cost of the system and the cost to maintain and support both the operating computer system and specific programming requirements are critical considerations in selecting an appropriate system.

Adequate performance. The system selected must be responsive and provide adequate computing speed, disk storage, and working memory space to address the needs of the surveillance program.

9.3 Hardware

Computer hardware. Individual work stations and overall processing platforms should be selected to handle the work of the surveillance program and allow simultaneous on-line use of the data by multiple users and the various software packages used by the staff. Systems speed, number of concurrent users, active memory, disk capacity, robustness, and compatibility are all important considerations.

Systems back-up hardware matched to size of system. Systems back-up strategies must be complemented by hardware of sufficient size and speed to generate systems back-up on a prescribed schedule without eroding systems performance.

Printers, printing capacity, and quality/variety. Hardware that permits printing in the volume required by the program and that will produce high-quality printed tables, charts, and reports is important. Printing capabilities may be required for high-volume printing of envelopes or other specialized printing. If a surveillance program has a large-scale follow-back, the ability to print in-house materials and mailings that carry names and addresses significantly enhances security of the information at relatively low cost.

Graphics and slide production capabilities. Hardware that can be used to develop Microsoft PowerPoint presentations or slides is important. Slide makers and LCD projectors should be available to surveillance program staff.

Communications hardware and links. The staff of the birth defects surveillance program must be able to send and receive e-mail and to access the Internet. Data collection through hospitals and use of data by staff must also be supported by appropriate computer communication systems.

Strategy for planned obsolescence. The hardware used by the surveillance program must be able to operate software and systems that are actively supported by the software or systems suppliers. Planning for replacement of existing hardware should be an ongoing process. This should ensure keeping pace with changing software and systems requirements, enabling staff to manage the surveillance database effectively.

9.4 Software

The basic software selected to run the surveillance database must provide the features required to meet programmatic needs. It must have the capacity and robustness to conduct required procedures, be compatible with other similar data management systems, and be supportable.

Data analysis software. Standard statistical software should be available to analyze surveillance data. Statistical packages must have a full range of capabilities for developing standard statistical tables (including counts and rates) and conducting more complex analyses (such as standard error calculations or observed-to-expected ratio estimates). In addition, the software must support the design of tables, as well as presentation features such as titles, footnotes, graphics and, potentially, mapping. (See also Chapter 8 on Statistical Methods).

Record locking and file locking. The data management system must provide for data security and confidentiality as well. Systems should be considered with confidentiality and security features that enhance the proper protection of data. Depending upon the types of direct database access various system users are permitted, the data management software may need to control data access at the level of the file, the record, and the individual variables. This may require various levels of file access, which could be handled by using data management software with these capabilities.

File security software. Software that can regulate access to file servers and to specific computer drives or computer files, and maintain various levels of file access rights, is essential for storage of data on a Local Area Network (LAN)-based or mainframe system. Staff should either manage the administrative features within this software, or these activities should be under their direct supervision.

Multi-user capability. Software used to access and manipulate the data may need to have multi-user capabilities, allowing access by multiple users during most, if not all, file management routines. The need for this capability will depend on staff size and the scope of the surveillance program.

Integrated and stand-alone utility programs. The database must be accessible to program staff. Software and skills needed to develop ad hoc and specialty software routines must be available, as necessary, to manage and maintain files or to conduct specialized analyses. Such custom routines may be required on an ad hoc or a routine basis.

Record linkage software. Software is needed that supports data linkage. This capability is essential to de-duplicate new report data and to link cases to corollary files, such as birth or death files. Record linkage capabilities are also essential to the conduct of cohort studies that can link cases to files of study subjects. The ability to link data, with a high degree of accuracy, is critical to data quality, to conducting basic surveillance functions, and to research. Some states have developed their own custom – designed programs to meet record linkage requirements (e.g., Colorado).

Linkage capability can take several forms, ranging from on-line case-by-case queries to electronic comparisons of large databases. Data can be linked electronically through either pre-programmed routines or ad hoc routines and can be based on deterministic or probabilistic linkage procedures. The specific strategy and approach used by a surveillance program will depend on its size, overall mission, and resources available.

- > **Deterministic record linkage** procedures, which involves the literal comparison of fields or columns within fields for exact matches, can be developed relatively easily and can be supported by most database management software. If this approach is used, it is essential to audit and refine the procedure painstakingly to ensure a high degree of matching accuracy.
- ➤ **Probabilistic linkage** bases record linkage decisions on determined probabilities that two records are likely matches. This technique generally is accepted as quite reliable when applied appropriately. However, it is dependent upon costly proprietary software packages that may not interface well with other data systems used by the surveillance program.

Regardless of the approach used, the results obtained through record linkage must be reviewed periodically for quality. The presence of unidentified duplicates within the case data and combining report data for different children into a single record are two obvious hazards of improper linkage. These false positive and false negative rates must be minimized by reviewing linkage quality regularly.

9.5 Process Standards

In this section we discuss the following aspects of process standards: inputs into the surveillance system (Section 9.5.1), instructions for reporting facilities on proper submission of data (9.5.2), procedures for initial review and query of submitted data (Section 9.5.3), procedures for receipt and logging of shipments (Section 9.5.4), and forms and batch control procedures (Section 9.5.5).

9.5.1 Inputs

Proper management of data within the surveillance program needs to begin through careful coordination with those providing the data and through following appropriate internal practices and procedures. These must be designed to promote accurate reporting and complete processing, ensuring a trackable system where processed data can be re-traced back through to the data originally submitted.

The procedures developed need to accommodate the various forms in which data are reported and the sources from which the data are derived. Data coming in to a surveillance program can vary widely with respect to the way they are transmitted and their content. This is true across states and within a state. The data can be provided in the following ways:

- ➤ Paper/electronic abstracts for reportable conditions
- ➤ Hospital discharge data
- Medicaid data
- > Early intervention program data
- > Data on services to children with special needs
- ➤ Birth and death record data
- ➤ Medical examiners reports

9.5.2 Instructions on Proper Submission of Data

Clear and concise instructions must be developed and distributed to all those involved in reporting cases to the surveillance program. Necessary components of these instructions include:

- Precise definitions of what constitutes a reportable condition/case
- > Item-by-item explanation of information to be reported
- > Timelines for reporting
- Acceptable reporting methods paper, electronic, File Transfer Protocol (FTP)
- ➤ How and where to ship reports
- > Procedures recommended to ensure secure shipment
- Procedures for handling corrections and updates to previous reports
- > Sample of abstract
- > Detail on electronic submissions, if appropriate
- > Definition of terms, as appropriate

Name of a contact person in case of questions

These instructions should be readily available to those with a 'need to know', should be prepared to minimize any anticipated potential misunderstandings, and should be updated routinely. Instructions need to be customized and targeted to specific data sources – such as laboratories, hospitals, physicians, or medical examiners – to reflect differences in what is expected from each.

Complete documentation of the receipt and preparation of case data from internal sources is also required. Examples include information from data systems for programs that provide specific information on children with reportable conditions. The nature of the data system and the schedule for providing or obtaining data, the format and technical specifications of the data all need to be documented. This is necessary to ensure coordination between the birth defects surveillance program and other data systems and/or sources.

9.5.3 Initial Review and Query

As reports are received, and during the intake process, it is important to establish procedures to screen incoming reports and data. These screens should be designed to avoid unnecessary work and to identify and resolve quickly any gross problems with the submission. These screening activities might result in a submission being returned and not processed. Other types of screening may occur at various points within intake processing of reports.

For paper reports, potential screens could include very basic things, such as examining the mailing for physical damage, proper addressing or, perhaps, tampering. Paper reports could be screened, as they are inventoried, to be sure they are completed adequately and that the case is truly reportable. If the paper report is primarily a case-finding tool, it might first be screened against cases in the database to determine whether it is new.

Pre-screening of data submissions should include checking for possible computer viruses, determining whether the file is readable and in an appropriate format and file structure, and establishing whether the count of records within the file is correct

9.5.4 Receipt and Logging of Shipments

As data are received in the form of paper reports or automated files, forms- and data-control procedures need to be followed. Procedures should be designed to ensure that all data submissions are processed. They must also provide a mechanism for rechecking the status of the surveillance program database to validate that all information has been processed properly and appropriately. This can be accomplished by developing a log to record receipt and processing of reports by facility. Such a log could contain basic information about each submission. This might include: date received, reporting facility, number of reports, date span for the reports, format of the reports, date prepared, report numbers assigned to the batch, file or batch name assigned to the data, and the date data processing was completed. Maintenance of a log serves as a control point for the data. It can also be designed to permit monitoring of the reporting status of individual facilities.

Depending upon the type of report being processed, other approaches may be appropriate to ensure completeness. For example, data that serve primarily for case finding may need to be screened first against the surveillance program's database to determine whether the report is for a new case, thereby permitting previously reported cases to be quickly dropped from the case-finding data.

9.5.5 Forms and Batch Control Procedures

For data control and tracking, it is important to use a systematic procedure to inventory and to identify unique reports. Classically this is accomplished for paper reports through use of a series of sequential numbers, with each form assigned a unique number. Forms are then organized sequentially into batches of manageable size. A similar procedure can be used for automated data submissions to assign each electronic record a unique identifier and to maintain a record of the file name assigned to each batch of reports submitted.

These procedures allow staff to locate a specific report easily and provide a mechanism for data inventory. Missing report numbers can be listed and resolved as report file completeness is evaluated. Data edits for each report can reference these numbers to identify and resolve any concerns with the data in that report.

9.6 Data Entry

The most basic aspect of developing a surveillance database is preparing an electronic file of reported information. While the proportion of paper case reports received varies widely across surveillance programs, each program must have reliable mechanisms for entering data from manual reports into an electronic file. In addition, many programs can provide reporting sources with software that can be used for submitting cases. Applying some simple concepts to the automation of information can help provide data files of consistent quality.

Interactive edits. Developing a process for capturing data destined to reach the surveillance provides an opportunity to build functional editing of entries into the operating procedures. This is especially effective when edits are used to question incoming data at the point where the patient's chart is available for review. Editing data at the point of origin is the most efficient method to ensure high quality.

Interactive edits can be very simple checks, such as ensuring that only numbers are entered into a numeric field, ensuring a date entered is a valid date, or preventing a required field from being left blank. More complex edits might involve providing links to a database of valid codes for diagnoses or procedures, editing for consistency across fields, or screening each case to determine if the child was reported previously.

In designing and developing interactive editing procedures, it is important that the objectives be kept in focus. A process is needed for producing high-quality electronic data efficiently and effectively. Interactive editing needs to be functional. It should be designed to screen for impossible or improbable entries. It must also be efficient, providing the operator with a clear explanation of the perceived problem and a ready mechanism for resolving the issue.

Verification. Verification procedures are another tool for controlling the quality of incoming data. The key data processing steps of information coding and the actual process of data entry are candidates for verification. Verifying data is an old and time-tested method of monitoring and controlling errors introduced into data through data processing procedures. These practices do not improve the quality of the reported data, but they do minimize degradation in data quality during data processing. The purest example of data entry verification is blindly re-keying previously entered data using software that compares the newly keyed data, key stroke by key stroke, to that entered earlier. Any discrepancies are identified and resolved by the verifier.

To verify data is to double check the data to ensure it is captured accurately. Verification procedures can take two basic forms, namely, independent and dependent verification. There are also two basic strategies relative to the scope of verification: verifying each incoming case or verifying a sample of cases. In *independent verification*, the verifier is not provided with the previous work and must essentially redo the work. The two versions are then compared and any discrepancies resolved. For *dependent verification*, the verifier has access to the original work and reviews the entered data, comparing it to the source document; in the case of data entry, essentially proof reading the work.

Focusing on verification as a tool for efficiently developing data files of consistent quality, verification can be developed incorporating the quality and skill of the processing staff with efficient methods for screening and resolving processing errors. As an example, dependent verification of all the diagnostic coding done by new staff might be done by experienced staff and continued until a "qualified" level of accuracy is consistently demonstrated. Once the new staff member has qualified, only sample independent verification might be done.

Information obtained through verification can provide important insights into staff training needs. These results can also ensure a consistency of understanding and interpretation across staff involved in data preparation, highlighting any inconsistencies.

Forms/record and batch controls. Since data arrive in a variety of forms and from numerous sources throughout the year, effective methods to inventory all incoming data are important. As a corollary to logging the receipt of data shipments, control of individual records is very important.

As reports – both paper and electronic – are received and early on in their processing, a report number needs to be assigned to each report to serve as its unique identifier. This identifier provides a ready mechanism to inventory the incoming reports and, later, the consolidated files of processed reports. This report identifier also enhances coordination of the work during later stages of file editing and processing.

There are a variety of schemes for assigning a report identifier. The most basic is a sequential number that begins with the year the report was received followed by a simple sequential number. By including record type/source codes within the prefix for the sequential number, the type of report or information source can be incorporated into this identifier. Such information is often useful in developing management information regarding database status.

A system for numbering data entry work files needs to be developed and employed to properly control and inventory work batches. Each work batch needs to be assigned a unique batch identifier. A log should be established to record the report identifier numbers within each batch. The log should include the date completed, the individual completing the batch, the individual verifying the batch, and the date the batch was processed into the surveillance program. This information will aid in assuring all reports are processed and in tracking down any discrepancies. Information in the log will help assess processing issues, such as timeliness and staff accountability.

Procedures appropriate for a variety of data inputs. It is important to map out the proper handling and intermeshing of data from each data source carefully to ensure data quality. As mentioned earlier, sources of information can vary widely, both in type and quality of data. In designing the data entry process, the form in which the incoming data are presented can create a need for customized procedures.

Tailoring the procedures to match the data source and data format can add to efficiency and enhance final data quality. These adjustments might take many forms, including facilitating data entry through customized data entry screens for certain report types. Specialized editing to match the data source and, perhaps, to screen for code conversion errors may be required. Some data sources might be considered primarily as sources of case ascertainment. The first processing step might be screening cases against the program's database to determine if the case has been reported previously.

Training/certification and instruction for data preparation. Program staff members involved in data collection and processing must have the skills required to accomplish their work accurately. The skills required vary across key functional activities, namely abstracting case data, coding the information, and entering the data. Data management and editing routines will not correct data quality problems that occur if staff members are not properly trained.

Surveillance programs need to have a strategy for training new staff that allows them to learn the new job; measures their understanding of the work; provides feedback on problems and progress; and determines, in some objective way, that the new staff member's work has reached an acceptable level of quality. Staff skills and the rigor with which work is reviewed will vary among surveillance programs. Whether a surveillance program utilizes active or passive case ascertainment influences the skills needed. Hiring staff with training and experience in health information management may prove important. By the very

nature of birth defects surveillance, there will always be a need to train new staff in a number of areas that are unique to the program and where it is not possible to hire experienced staff. There must be a strategy to ensure that staff assigned to a task have the skills the task requires.

As a component of continuous training, detailed manuals are needed that document procedures and serve as a reference source for staff. Staff should be encouraged to refer to these manuals and to identify errors, inconsistencies, and misinterpreted sections. Updating these guides periodically ensures that the manuals/instructions remain functional and current and able to serve as training guides for new staff.

Future editions of *The Surveillance Guidelines* will address training issues for surveillance programs in greater depth.

Input file processing functions. The management of data quality within electronic birth defects data files is important as well. The procedures and processes for handling the quality of processed electronic data are similar to those used for paper reports. The tools available to a birth defects surveillance program are basic data processing and management practices that are not unique to these types of data. Electronic data files readily lend themselves to editing and clean-up. Standard computer routines can be used to screen files for obvious errors or inconsistencies, to spot problems with the data efficiently, to summarize findings, and to organize results in ways that allow the efficient correction of any errors.

Key components of input batch processing are outlined to provide an inventory of the tools available for functional data quality control. The combination of practices employed by a given surveillance program needs to match the methods and procedures used for data file development.

Editing. Development of data editing procedures is a standard activity in any database development effort. As with interactive editing during data entry, computer routines can be developed to identify a variety of data problems. Standard edits often include:

- > Field range checks
- Report number range checks to identify missing records
- ➤ Inter-item consistency checks
- > Field validity checks
- ➤ Code validation through table look-up, i.e., diagnostic or procedures code tables
- Consistency across multiple reports for the same case
- ➤ Hard versus soft edits and use of edit flags

The organization of the results from edits requires the same care in design as do the edit criteria. The results of an edit run need to be organized to make error resolution and file correction as efficient as practical.

Tracking information. As potential problems with data are identified, it may be necessary to ask the reporting source for clarification or for additional information. A basic procedure to monitor outstanding requests for clarification or correction should be used.

Printed case abstract. In conjunction with efforts to correspond with staff at the data source about reports, a ready mechanism to print an abstract of a report can improve the effectiveness of communications and may enable correction of other errors in a report that cannot be identified by the edit routines. The capability to print a case summary easily can prove useful for multiple purposes.

Error correction. It is important to have effective and efficient procedures for error correction. Reports that identify edit exceptions can be linked to the edit results to pull up rapidly, or to *queue*, the records needing attention. A well-designed process can minimize the potential for introduction of errors in the course of record correction.

Case-by-case and multi-record correction. Mechanisms to correct records one at a time are important. The capability to update multiple records simultaneously can also be useful. When used judiciously, multi-record correction can save time and reduce the potential for error.

Add/delete. The capability to delete spurious or redundant records can prove to be very useful.

Linkage and assignment of case identifier. As input files are processed and screened for duplication, a system for uniquely identifying each case is necessary. While a program may choose to number and retain all reports received, it is critical that a specific child's reports all have the same case identifier. This is necessary for record and file linkage. In a program where data are consolidated immediately, an identifier for each child is still a critical component of the system. A mechanism for assigning identifiers to newly reported cases is necessary. In the case of electronic submissions, this process should be automated.

Facility reports. Summaries of data quality relative to screening and editing of incoming reports is important for maintaining an accurate picture of the quality of submitted data. Summary reports that permit the tracking of report quality over time and across facilities can be designed. Such information is very useful in identifying facilities that are candidates for data quality reviews and/or in-service training. These reports can complement efforts to work with facility staff to correct any persistent problems.

9.7 Record Linkage

The proper operation of any birth defects surveillance program depends on developing and following procedures for efficient and effective record linkage. These procedures should be developed carefully. The accuracy of the procedures used to link individual case reports needs to be measured and monitored. Instances of the same child being in the database as different children and different children being presumed to be the same child must be estimated. Developing and monitoring linkage procedures carefully is as crucial for programs that manually search for potential matches as it is for those that use electronic linkage.

Not only is it important to link incoming reports accurately to the historic file to locate previously reported cases, but the ability to link to other databases is also essential. Procedures need to be tailored and evaluated specifically for each type of linkage. Goals for linkage completeness that reflect these expectations need to be established.

Linking birth defects case data with files from other sources may be done to meet a number of objectives. These include:

- Deleting duplicate data
- Case-finding
- ➤ Augmenting the information available for a case
- > Conducting special studies or program evaluations

The level of precision and efficiency that can be expected from a matching process are functions of several factors. Key among these are:

- > Ouality of data within the files to be linked
- Number of fields common to both databases
- ➤ Logic employed to compare the files
- > Time available to review and assess each link

The matching strategy developed should maximize the results and minimize the resources employed to obtain those results. Estimating the level of precision for any linkage procedure can be used to assess the advisability of revision. These estimates are also important for evaluating the suitability of using the linked data for specific purposes.

With respect to assembling the required data on each case, linkages to birth certificate files and death certificate files are extremely important. These sources can provide the surveillance program with valuable information on each case. For example, linkage to the birth certificate file has the added benefit of identifying reports for a single child that may not have been linked properly during the processing of incoming data. Linkage to birth and death records can also provide the ability to track changes to a child's name over time. This can assist in collating data on a single child that might otherwise be treated as distinct cases. In some jurisdictions, access to this kind of information will depend on legally prescribed restrictions.

9.8 Record Consolidation

When multiple reports are received on the same case, differences can be expected in some of the information across reports. By developing a summary of the information on each case, consolidating the information across reports into a single summary, the information about the case can be enhanced. A number of issues must be addressed in any information consolidation effort. The categories of information that could be consolidated or summarized, and the key issues relative to summarization, include (1) demographics and identifiers and (2) diagnostics.

Demographics and identifiers. Most demographic items are constants and do not change with the age of the child. These include date of birth, race or ancestry, mother's age. Updating missing data fields using data from subsequent reports is generally appropriate. Conflicts across reports for these fields can be difficult to resolve, but may be predicated on the source of the data, prioritizing data from specific files or facilities. Changes can be expected to occur over time in identifier fields such as name, parents' names, and address. Selection of the appropriate data to be included in a summary needs to be based on the purpose of the summary. For example, data for referral or outreach efforts need to be current, while data for auditing birthing hospital records should represent information at birth.

Diagnostics. As multiple reports for a child are received, collecting diagnostic information across all reports can result in significant redundancy. When the same diagnoses are reported repeatedly, this redundancy is simple to manage. As diagnostic data change across reports, there are three possible causes. Each of these raises specific issues relative to proper management:

- New conditions being diagnosed. Newly diagnosed conditions clearly must be included in any summary for the child.
- ➤ Previously diagnosed conditions reported with greater or lesser specificity. Redundancies in diagnostic codes caused by differences in specificity can be problematic. In the absence of accepted guidelines for doing so, to eliminate redundancies (increasingly specific diagnostic codes) using intuitive logic can be problematic. The logic must be thought through clearly, with the intended use of the resulting summary in mind.
- ➤ Actual changes to a previous diagnosis. Changing a diagnosis can reflect a revision based on better information or an alternative diagnosis, for example, a difference of opinion. A key problem with such changes is the need to differentiate a changed diagnosis from a condition that has been newly diagnosed. Having an effective mechanism in place for facilities to report corrections to diagnostic information can help reduce confusion in interpreting subsequent reports. Making changes to diagnostic data should be done carefully and through close coordination with facility staff. Some programs may prefer not to change original information, but rather flag it as inaccurate or no longer valid. In this way, the integrity of the database remains intact: inaccurate information is not "counted", data quality evaluations can be conducted, and new or confirmed information is accepted. This is especially useful when comparing reported information with the results of medical records review by surveillance staff.

Procedures data. Redundancy can be expected in data reported for procedures, since it is not uncommon for a child to have some treatment or corrective procedures performed multiple times. Therefore, such data can be consolidated reliably only if the date each procedure was performed is reported, or available through medical record review, to identify specific procedures by date received.

9.9 Feedback to Data Sources/Abstractors

When data problems are identified during report processing, it is important to communicate those problems to staff at the data source. Facilities and/or individuals providing data should be interested in learning of problems the surveillance system encounters with respect to the completeness and accuracy of their data. With passive reporting systems, communicating errors, resolving inconsistencies, and reviewing apparent discrepancies represent effective feedback mechanisms. An efficient mechanism is needed to provide feedback on problems, although the necessary corrections to the information may seem obvious to surveillance program staff. Staff of the reporting facility will benefit from the feedback and may need to correct the information within their own records.

9.10 Security

Many of the data assembled by a birth defects surveillance program are extremely sensitive. For this reason, a program must initiate and maintain a comprehensive strategy for data security that ensures data are protected from improper access or inappropriate use. Developing a security plan that establishes and demonstrates a commitment to data protection is essential in reaching the program's long-term goals and objectives.

9.10.1 Personnel Issues

Four aspects of security management fall under the category of personnel issues.

- ➤ Hiring practices. Attention needs to be paid in selecting new staff members to screening candidates to ensure they can be relied upon to handle confidential data appropriately. A work history that includes responsibly handling confidential data is an example of desirable experience. It is important to request and check references for all prospective employees. If possible, security background checks should be conducted prior to making a hiring decision.
- ➤ Written procedures on security and access. New employees need to be informed clearly of the procedures regarding appropriate access to and use of data, particularly any files that include personal identifiers. Written materials that describe the nature of the data and the rules and policies relative to data handling must be reviewed with employees. These materials must cover all aspects of employees' actions for which they are accountable. These materials need to be discussed with each employee to ensure that the employee has every opportunity to ask questions so that they understand the policies explicitly. A written policy on the release of identifiable or potentially identifiable data must be included. It is essential that all aspects of data release be identified within the policy, along with who has the authority to authorize a release. Such a policy must include the "business activities" of returning data diskettes and corresponding on data editing problems with data providers, as well as release of identifiable data for research use or in conjunction with child-find referral activities.
- > Security and confidentiality agreement/oath. Each employee who has access to the program's data must sign a confidentiality pledge. The pledge should be in the form of a comprehensive statement that outlines the confidentiality policy in broad terms. In addition, this document should include a statement that the employee understands the confidentiality policies and the potential consequences for violating these policies. Finally, the document must include an oath on the part of the employee that they will abide by these policies. As significant changes to the confidentiality policies are made, each employee must sign a new pledge that reflects the new policies.
- Disciplinary policy. Whenever there is an allegation of mishandling confidential data or where unauthorized access is suspected, the incident must be investigated. Such an investigation must be conducted carefully and in a manner consistent with existing employment laws and personnel practices. Appropriate disciplinary action must be taken if it is established that an employee has violated the confidentiality policy. Any deliberate violation of policy that results in the inappropriate release of confidential data should be grounds for dismissal and for potential criminal action, depending upon the law governing these data.

9.10.2 Transportation and Information Handling

Basic security concepts that should be considered relative to shipping and handling information are listed in this section. Standard office practices and procedures for handling materials that include confidential data need to be developed and followed. These are necessary to avoid problems due to inappropriate or inadvertent access to these data.

The privacy regulations developed as part of the Health Insurance Portability and Accountability Act, or HIPAA, place significant responsibilities on hospitals, physicians, and others to properly safeguard confidential data on their patients. These regulations place strict procedural standards on health care facilities, heightening concerns about patient privacy held by health care facilities and providers. It is important to adopt information exchange practices with data sources that do not create a potential liability under the provisions of HIPAA (see Chapter 2 on Legislation). For example, common and efficient methods for exchanging information, such as fax or e-mail, need to be avoided or used with great attention to appropriate security. This is because faxed images can be intercepted and printed, can be inadvertently sent to the wrong fax or to a fax that is unattended or otherwise not secure. E-mail shares all these problems in addition to the fact that e-mailed materials will become part of the e-mail back-up systems and copies of sensitive materials will become interspersed with other documents that may well be public information. The existence of these back-up files means a loss of control over the data and access to the data. The problem of potentially intercepting e-mail only compounds this problem.

With these thoughts in mind, key considerations relative to good data handling and transporting practices are provided below.

- Instructions to data sources for addressing and shipping of incoming reports and information. All facilities and individuals who ship abstracts or data to the surveillance program must be provided with current and precise address information. Data sources should be encouraged to ship data in a secure manner where chain of custody signatures are required, such as certified mail or FedEx. If a shipment is received that was misaddressed, the data source reporting the cases should be contacted promptly by telephone, with a follow-up letter, and be advised of the correct addressing of shipments. In addition, standard practice should include prompt acknowledgement of shipment receipt. As staff at the data source learn to expect an acknowledgement of each data shipment, failure to receive an acknowledgement will alert them to the possibility that a shipment has been lost or delayed.
- ➤ Use of fax or e-mail for forwarding or receiving sensitive data is not advisable. These methods of transmission should not be considered secure unless the sensitive information is encrypted and password protected. Fax machines that both send and receive such materials need to be attended during transmission.
- Managing the work station. Employees need to be trained to manage their desktops. Confidential materials should not be on a desk if they are not being used actively and should not be left unattended during breaks or lunch periods. Similarly, passwords should be required for staff access to any personal computer that holds confidential data or that allows access to confidential data through a network or other computer connection. Such equipment should not be left unattended with connections in place that would permit unauthorized access. Care must be used in displaying confidential data on the computer monitor in order to ensure that persons who do not have authorized access cannot read them. All materials must be filed properly and locked when not in use. Following these common sense practices reduces the possibility of inappropriate access and should be applied conscientiously to the desk, the personal computer, and the files assigned to each staff member.

- Physical access to abstracts, other documents. Limiting direct physical access to files and other materials that include confidential data is a basic step in reducing the likelihood of someone seeing information to which they are not privy. The design and layout of offices can be done in a way that enhances staff members' ability to carry out their responsibilities without exposing confidential material to others. Careful planning in this regard, combined with good desktop management practice, will minimize inadvertent access.
- ➤ Procedures and furnishings to lock up documents and diskettes. Employees must be provided with the office furnishings needed to adequately secure documents. Locking desks and locking file cabinets are essential, with thought given to assignment and management of keys for these locks and the organized storage of extra keys.
- ➤ Procedures for shipping reports and information from the program. Program staff need to follow secure practices, as when they send any confidential materials to data sources. It is essential that such shipments are addressed properly, and the address should be confirmed if there is any doubt about its correctness. Materials should be shipped using a method/carrier that obtains a signature to verify receipt. Confidential data should not be shipped through e-mail or FTP unless the security of the connection is ensured or an adequate encryption technique is used to disguise the data
- Shredding and destruction. Considerable care must be taken to avoid any potential for disclosure of data when confidential material is discarded. Employees must be conscious of the need for care when discarding any program-related materials that include identifiers or that would be considered confidential. Computer listings, correspondence, and other materials need to be screened to be sure that confidential data are handled appropriately. Staff should be provided with access to a shredder, and paper abstracts or printouts with confidential data should be shredded promptly. Any large volumes of confidential materials that need to be disposed of must be destroyed in a secure way.
 - Similar standard precautions must be established for computer storage devices. Diskettes should be reformatted, rather than simply deleting files. As hard drives on personal computers are replaced, the old drive must be reformatted or any data remnants otherwise destroyed, for example, by storing them between strong magnets for a period of time.
- > Transportation of data. When staff members are in the field, all confidential data must be carefully safeguarded. Documents should be transported in locked brief cases or otherwise protected. The security of portable computers must be ensured. Confidential materials must be kept locked in the vehicle trunk while traveling. During overnight stays these materials should be removed from the vehicle and placed in a hotel room rather than left in a vehicle overnight.

9.10.3 Physical Security

Physical features of the worksite can enhance information security significantly. There are two specific ways the facility housing a surveillance program can maximize security:

- Restrict physical access to the work area. To the degree possible, access to the surveillance program's work area should be controlled. Ideally, it should be isolated with a card entry access system. Reducing or eliminating travel into and through the work area translates directly into reducing or eliminating opportunity for inappropriate data access.
- After-hours security. At a minimum, the office area must be locked securely after working hours. Ideally, the office area should be protected against unauthorized access through use of an alarm system that includes motion detectors and that is monitored centrally and continuously. If possible, no janitorial services should be carried on after hours.

Periodic maintenance work should generally not be conducted after hours unless surveillance staff are alerted and have an opportunity to take any and all extra precautions to ensure appropriate security of the data

9.10.4 Computer Security

Proper data security requires a comprehensive approach to computer security. A number of key aspects to any plan designed to protect electronic data files are listed below.

- ➤ User ID and password. A system for unique user IDs and passwords is a cornerstone of computer network security. Staff should not be allowed to share ID and password information. Departing employees must be deleted from the system promptly. Periodic outdating and changing of passwords should be standard. Employees need to understand the importance of these activities and know that their personal login is critical to protect. This is because activities on the system will be traceable to the user's ID and password.
- ➤ Virus scan current. In receiving electronic data, it is essential that diskettes and other electronic files be scanned for viruses prior to loading onto the personal computer or the network. A comprehensive and continuously updated virus scanning package should be used for this purpose.
- Control of user access to data. Careful management of user rights to the network or other computer system can significantly enhance data security. It is important to minimize to the extent possible unnecessary access to files. Steps must be taken to decrease or eliminate both potential misuse of data and inadvertent damage or destruction of data that are accessed inappropriately. Planning the architecture for data storage can complement limiting access to the various data files and greatly enhance security in the process. Much like user IDs and passwords, this level of security must be continuously maintained, with access modified as staff work assignments change over time.
- > Discarding of old personal computers, hard drives. There are a number of special considerations regarding the security of electronic files. Simply deleting a file from a hard drive or diskette does not actually erase the data. This problem is not always properly addressed as old computer equipment is swapped out or discarded. There must be procedures developed for disposal of computer storage devices that ensure none of the data are recoverable.

9.10.5 Policy on Release of Data

Written procedures must be established that describe the proper mechanisms for release of information from the surveillance program. Written procedures are necessary to provide surveillance staff with a clear understanding of proper data handling and release. The process for obtaining approval for access to the data, and authorization for release of the information, must be described in detail. There must be no confusion among the staff on this critical topic.

Confidential data release procedures should include the specific practices required for proper preparation of tabular statistical data, as well as de-identified micro data files. These micro data files must be designed to guard against inadvertent disclosure of confidential data. The procedures must delineate clearly the approval process that governs and regulates release of identifiable or potentially identifiable data. Issues related to sending identifiable information to data sources and to other sources of information about cases of birth defects must be covered. Providing access to confidential information for research purposes must be discussed, describing the types of research projects that may gain access to these data and the system's process for reviewing and approving such projects. Finally, the conditions under which the information can be used for administrative purposes must be covered. This should include using the data to ensure that children and families are referred appropriately for needed services, if this is part of the surveillance system's objectives.

Chapter 10 Data Collaboration and Dissemination through the NBDPN

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10.1 Data Collaboration and Dissemination through the NBDPN

With the support and collaboration of the Centers for Disease Control and Prevention, the National Birth Defects Prevention Network (NBDPN) collects, analyzes, and disseminates state- and population-based birth defects surveillance data. In this way, the NBDPN plays an active role in turning data from throughout the US into useful information and encourages the use of birth defect data for decisions regarding health services planning, such as secondary disabilities prevention and referral to services.

The NBDPN State Data Committee coordinates the procedures and processes required in this effort. One of this committee's collaborative activities is the publication of an annual report of birth defects programs. The report includes detailed descriptions of the individual state birth defects surveillance programs, tables of data submitted by the participating states, and selected data analyses. The report includes diagnoses of interest, information regarding the format for submitting data for the annual report, and the criteria for the state directory that describes each birth defects surveillance program.

The NBDPN State Data Committee also discusses issues related to data suppression, confidence intervals, statistical analysis, and presentation of data, which are often of concern to the states. Because of the committee members' technical expertise in working with state data, the committee also has a role in evaluating the feasibility and merits of NBDPN participation in other data projects. Any use of data aggregated under the auspices of the NBDPN, including data projects and ad hoc studies, must be approved by the NBDPN Executive Committee.

All decisions regarding privacy, security, and confidentiality issues related to releasing or submitting surveillance data from individual programs to the NBDPN are handled at the state level.

Interested parties are invited to refer to the NBDPN website (at http://www.nbdpn.org) for contact information relating to the State Data Committee and for instructions on how to submit data to the NBDPN for the annual report or for ongoing special projects.



Guidelines for Conducting Birth Defects Surveillance

Chapter 11 Data Presentation

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Comments and suggestions on this document are welcome. Submit comments to the Surveillance Guidelines and Standards Committee via e-mail at nbdpn@cdc.gov.

You may also contact a member of the NBDPN Executive Committee by accessing http://www.nbdpn.org and then selecting Network Officers and Committees.

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11.1 Using Data for Decision-Making

This chapter focuses on the fundamentals of data presentation for a birth defects surveillance program. A birth defects research program will have needs that go beyond what is addressed in this chapter. Readers are referred to the references and technical appendices in this chapter for additional information. The reader may also wish to refer to Chapter 8 (Statistical Methods) of The Surveillance Guidelines for more in-depth treatment of some of the topics touched upon in this chapter. Finally, the Members Only section of the National Birth Defects Prevention Network (NBDPN) website will be posting materials on more advanced aspects of data presentation as they become available.

Collecting data for data's sake wastes precious resources. There is no good reason to collect data unless we intend to use them, generally to *inform* someone in a position to *do* something about the story our data tell.

Surveillance data in particular are intended for use in accomplishing the purposes and objectives of the surveillance program. In Chapter 1 of *The Surveillance Guidelines* we discussed the five major purposes of birth defects surveillance and their related objectives, as presented below.

- ➤ Epidemiologic. Epidemiologic objectives include developing timely baseline birth defects rates, monitoring trends and relationships to environmental factors, performing cluster investigations, and providing a basis for ecologic and etiologic studies
- ➤ Planning and prevention. Planning and prevention objectives include providing data for services planning, providing a basis for prevention strategies, and evaluating the efficacy of preventive services and programs.
- Educational and social. Educational and social objectives include informing the public about public health importance, informing parents about resources and care facilities, providing data for studies of economic impact, and providing data for follow-up studies of long-term effects.
- ➤ Healthcare and human services. Healthcare and human services objectives include referring children to services and resources and evaluating services utilization.
- *Clinical.* A clinical objective is providing the basis for clinical research.

Of course, not all surveillance programs pursue all of these purposes and objectives, but every program pursues some combination of them, and all collect data as a means to achieve them.

In order to fulfill the objectives of a birth defect surveillance program in all of these core areas, data must be collected in a complete, accurate, and timely manner. They must also be processed and interpreted in a way that ensures the availability of useful information to those with the responsibility to carry out specific activities that meet the program's objectives. Under some circumstances, this is relatively straightforward. For example, if a programmatic objective is to connect babies with specific birth defects and their families with appropriate medical and social services, then data collected on diagnosis and parent contact information immediately provide the information needed to initiate an appropriate referral. Frequently, however, there is a need to aggregate, analyze, and interpret data and subsequently present the resulting information to a variety of partners capable of taking necessary action. It is this latter more complex process that is the focus of this chapter.

Chapter 11 11-1 Data Presentation

11.1.1 The Data-to-Action Continuum

Yet data are, after all, only data. How is it that the data so carefully collected by surveillance program staff are transformed into the many different kinds of actions necessary to achieve their programmatic objectives?

There are two points to consider in answering this question. First, surveillance staff clearly cannot accomplish all of these important objectives without the help of their partners. Second, the transformation of data into action is not a discrete one-time occurrence—such as standing up with your slide presentation in front of a live audience—but rather a complex process involving extended collaboration between surveillance staff and their partners over time. To be sure, it is through presenting data in a clear manner in response to expressed interests of a particular "audience" and in support of an actionable message that this transformation begins to occur. Yet we need to bear in mind that, while the data presentation theories and skills discussed in this chapter can be mobilized in aid of this transformation, they are in fact only one aspect of the larger collaborative process that transforms data into action.

We can conceptualize this transformation as having four stages (see Figure 11.1, the Data-to-Action Matrix), with surveillance program staff and their partners closely involved in each one. Sources are abstracted to obtain *data*. Data are analyzed and interpreted to obtain *information*. Information is communicated to develop *knowledge*. And knowledge is used to inform *action*. Data presentation, then, is one of several skills that support this process, as we convey information to our program partners in order to generate the knowledge needed to embark on actions that meet our shared objectives.

11.1.2 Products of the Data-to-Action Transformation

Figure 11.1 suggests that each stage of the data-to-action transformation results in a distinct "product": data (Stage 1), information (Stage 2), knowledge (Stage 3), and action (Stage 4).

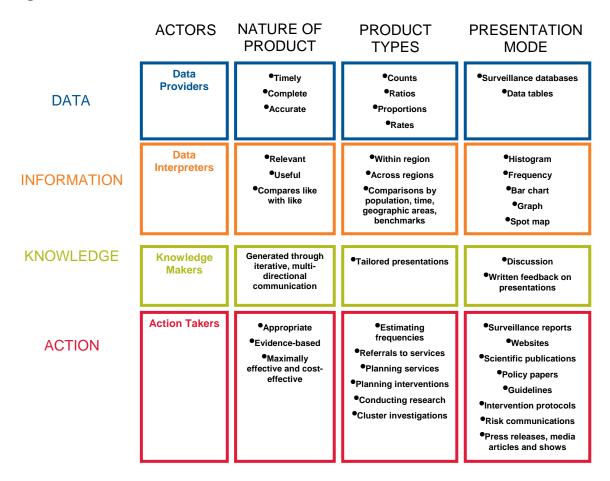
Let's take a moment to clarify these terms. While this chapter is entitled "Data Presentation," we are not really talking about presenting *data*, but rather about presenting the *information* generated from data in the expectation of building *knowledge* for ourselves and our partners. Although the terms 'data' and 'information' and even 'knowledge' are often used somewhat interchangeably, there are important distinctions between them.

Simply put, the purpose of data is to record "something" and the purpose of information is to build knowledge. Data (from the Latin meaning "something given") consist of raw facts or unedited stimuli. They are based on the symbolic recording of something, such as numbers, facts, and figures. Data provide a foundation for and can be developed into information, but they must be combined and integrated with other data before they become useful.

While information includes data, data do not necessarily include information. Information is data with semantic association and is the result of processing, manipulating, and organizing data in a way that adds to the knowledge of the receiver. When augmented by meaning or interpretation, data become information. It is the information developed from data that provides answers to our questions and those of our partners about birth defects, thereby increasing our knowledge.

Chapter 11 11-2 Data Presentation

Figure 11.1 Data-to-Action Matrix

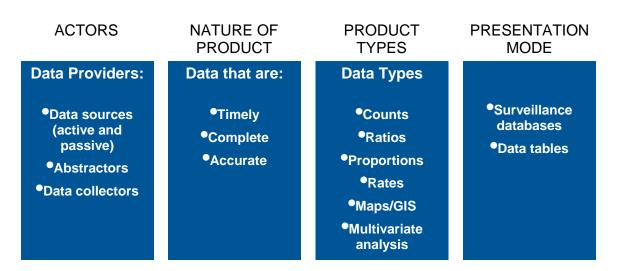


From the perspective of this chapter, our goal is to obtain data from a data provider (Stage 1–Data Provision), analyze and interpret it so that it becomes information (Stage 2–Data to Information), and then present it to and discuss it with one or more potential action takers so that it becomes knowledge that can be used to meet programmatic objectives (Stage 3–Information to Knowledge). It is then the action takers' responsibility to see that the new knowledge is used to meet the specific objectives of the program for which it has been collected and for which it is relevant (Stage 4–Knowledge to Action).

This chapter discusses each of the stages in the model in turn. We spend less time on the first and fourth stages, as both are thoroughly discussed elsewhere in *The Surveillance Guidelines*. Stage 4 (Knowledge to Action) is discussed further in Section 1.4 of *The Surveillance Guidelines* (Uses of Surveillance-based Birth Defects Data), and most of the rest of *The Surveillance Guidelines* address Stage 1 (Data Provision).

Chapter 11 11-3 Data Presentation

11.2 Stage 1 – Data Provision



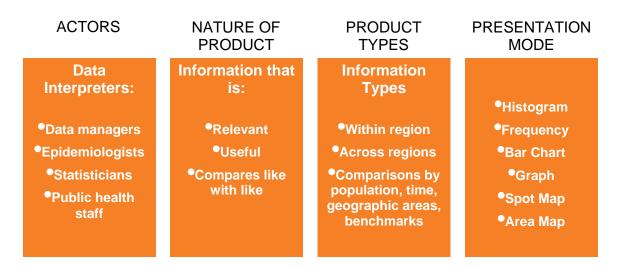
With respect to the ability of surveillance data to provide useful information, the old axiom from statistics "garbage in, garbage out" holds true. Before a surveillance staff member can begin to think about how to present data, knowing what data to present and feeling confident that the data are accurate and reliable is paramount.

In fact, the value provided by the information developed using birth defects surveillance data depends heavily on the quality of those data and the completeness and accuracy with which they are collected. The majority of the technical content of *The Surveillance Guidelines* is directed toward helping to ensure completeness and accuracy of the data that are collected and their resulting validity.

From an epidemiologic perspective, when we refer to *data validity* we are concerned with whether the data regarding cases in a study or surveillance program accurately reflect the numbers and characteristics of the cases that occur and that are eligible for inclusion in the data set. When we are attempting to determine or measure the occurrence of birth defects in a population, it is essential that we include all of the cases that meet the established case definition (completeness). For cross-sectional or case-control studies, while completeness is important, in the absence of including all cases validity is driven by whether the cases that are included accurately reflect all the cases that occurred in the study population (population at risk) with respect to epidemiologic variables related to characteristics of person, place, and time.

In the next section we discuss some of the analytical and interpretative issues involved in turning surveillance data into information.

11.3 Stage 2 – From Data to Information



In *The Surveillance Guidelines*, we follow the Centers for Disease Control and Prevention (CDC) definition of *surveillance* as established in Chapter 1. The key themes of the CDC definition of surveillance are the integration of data collection, analysis, interpretation, dissemination, and application. It is in moving from analysis to interpretation that data are converted to information.

Some aspects of birth defects lead to potential confusion or ambiguity in reporting information about them and their distribution. In this section we discuss a number of analytical and interpretive issues that should be considered when developing and presenting birth defects data. These include:

- Providing contextual information (person, place, and time)
- Missing or unknown data
- > Importance of comparison
- ➤ Approaches to measuring occurrence (prevalence versus incidence)
- ➤ Level of focus (which in part arises from the complex etiology and comorbid nature of many birth defects)
- ➤ Risk factors and the importance of timing with respect to exposures
- ➤ Privacy and data suppression (see also Appendix 11.1)
- ➤ Using Geographic Information Systems (see also Appendix 11.2)

11.3.1 Providing Contextual Information—Person, Place, and Time

When presenting data, it is useful to consider the key epidemiologic constructs of *person*, *place*, and *time*. What population is reflected in the data? From what location were data collected? And on what time period are the data based? One must be able to accurately and precisely answer these questions for the findings to be relevant. For example, a presentation may report very interesting results based on a sample that was collected in a very disorganized and biased manner, making it impossible to define exactly what

population is reflected in the data set. Unfortunately, these results would be of limited value because it is impossible to define to whom the findings are relevant.

Similarly, whenever variation by person, place, or time occurs, analyses should examine possible differences or trends. If a sample includes multiple ethnic groups, are there differences between these groups? Or if data were collected over a decade, were trends seen over time? A presentation should acknowledge that such trends were examined and differences reported if observed.

In the actual presentation, it is often useful to present data grouped on the basis of person, place, and time. When doing so, it is important to be mindful of widely accepted groupings inside or outside your organization. For example, *person* characteristics such as age, race, and ethnicity can be grouped based on Office of Management and Budget classifications. Audience members will be familiar with such groupings and, more importantly, they will be better able to relate the findings to their own data based upon these common groupings than they would if the presenter organized the data in some idiosyncratic manner. Similarly, *place* can be presented in a variety of ways, including aggregating based on town, county, zip code, or census tract.

Information collected over *time* can lead to more complex issues, such as the decision to report raw curves or a moving average. The complexity of time-varying data requires that one be clear on both the time period and method used in presenting such information.

11.3.2 Missing or Unknown Data

An aspect of data presentation often overlooked is the importance of providing information about the extent of missing/unknown data for study variables. Information that is missing or unknown can be just as important to understanding results as is the available information. This is especially true when the amount of missing information is more than minimal. Missing or unknown information can be reported in such data displays as tables, histograms, and pie charts by including a category labeled 'unknown' (e.g., maternal age \leq 34, maternal age 35+, and maternal age unknown). If the way the information is being presented does not allow for a row/column/line/bar/slice to be designated as 'unknown', a footnote should be added to the data display informing the audience about the extent of the unknown data. Maps based on geocoded data, for example, could add a footnote with the "percentage of data that was not geocoded" to the geographic resolution presented.

11.3.3 The Importance of Comparison

Epidemiologic data tend to be numeric and presented either as counts, ratios, proportions, or rates. In addition they are usually presented as information specific to the epidemiologic parameters of person, place, and time. Information presented in this manner provides a way of making meaningful comparisons between different populations and different periods of time. Note: the points made below are of particular importance when one will be comparing data collected at different levels (local, state, regional, national) or by different programs.

Fundamental to epidemiology are the principles of comparisons:

- ➤ Between areas/populations
- ➤ Within an area/population over time

These comparisons often involve consideration of epidemiologic variables such as sex, plurality, race/ethnicity, pregnancy outcome, maternal age, etc. Comparisons are also usually of some measure of

occurrence, in the case of birth defects *prevalence* of a specific malformation or groups of malformations. Increasingly, there is interest in making comparisons between some health status indicator at the local or state level and a benchmark, such as a Healthy People 2010 objective or an agency-developed objective.

For comparisons to convey useful information, it is essential that **like be compared with like**. When comparative data are presented, the audience must know if this holds. In terms of birth defects data, there are at least four points that need to be clearly established if meaningful comparisons are to be made:

- ➤ What is being counted? Are the outcomes—case definitions—comparable? (see Chapter 3)
- ➤ How are cases ascertained? Were similar methods of case ascertainment used? (see Chapter 6)
- Are the pregnancy outcomes from which the cases were ascertained comparable?
- ➤ Are comparable measures used to summarize data?

Each of these is worth considering with respect to the information that can be provided based on surveillance data.

What Is Being Counted?

Comparability of outcomes revolves around disease coding, classification, and the aggregation of cases. At the most general level, if we refer to "the occurrence of birth defects," we need to be clear about what is included in that term. In the past, birth defects were usually considered to be synonymous with congenital malformations and referred to diagnoses with ICD-8 and ICD-9 codes 740.0 to 759.9. Some surveillance programs, however, may follow the much more general March of Dimes definition of birth defects that includes metabolic and functional abnormalities as well. When comparing data between programs that use different definitions of the term 'birth defects', there are likely to be sufficient differences between what programs are counting as to make comparisons difficult, if not meaningless.

Even programs that use the same definition for the term 'birth defects' may vary in terms of what they include (and count) under a specific group of birth defects. One example of this relates to the reporting of studies of neural tube defects. In the past it was common to see reference to the occurrence of "central nervous system (CNS) malformations." Anencephaly and spina bifida might make up the majority of the cases, but cases of hydrocephaly and microcephaly would often be included as well. Clearly, comparing the results of a study that reported on the occurrence of all CNS malformations with one that consisted only of cases of anencephaly and spina bifida would be inappropriate.

Programs may also differ in the ways they define a specific birth defect. For example, most surveillance programs do not include preterm babies with atrial septal defects as cases. The Metropolitan Atlanta Congenital Defects Program (MACDP), for one, does not include infants of less than 36 weeks gestation at delivery among their reported cases of this defect (Correa et al., 2007). Therefore, if a program does not establish a gestational age criterion for atrial septal defect as part of the case definition, then comparison of their prevalence data with those of MACDP would be misleading.

How Are Cases Ascertained?

The second key aspect to data comparability relates to how the surveillance program ascertains cases. For example, some have expressed concern that surveillance programs relying on the reporting of cases by hospitals (passive case ascertainment) may identify a smaller percentage of the true cases that occur than will programs that send abstractors from their staff out to hospitals to actively search records for potential cases (active case ascertainment). Such differences may be more perceived than real, depending on the individual surveillance programs involved.

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Perhaps a better example of potential differences in completeness of ascertainment based on methods of case identification would be an attempt to compare data from a program that identifies cases only from vital records (birth and fetal death certificates) with data from a program that identifies cases based on medical record review. Several studies have identified serious problems with under-reporting of malformations on vital records (Watkins et al., 1996).

Are the Pregnancy Outcomes from Which Cases Were Ascertained Comparable?

Another issue with respect to comparisons relates to the populations from which cases are identified. While some surveillance programs are able to identify prenatally diagnosed cases that result in pregnancy termination and include them in their numerator, many are not. This difference is particularly important for defects such as anencephaly and spina bifida, which are being diagnosed prenatally with increasing frequency. In one of the first studies conducted by the NBDPN, prevalence data over time were presented separately for programs that did (9 states) and did not (13 states) ascertain prenatally diagnosed and electively terminated pregnancies where a fetus with anencephaly or spina bifida was identified (Williams et al., 2002b). Figures included in this paper clearly show the potential effects of inappropriately comparing prevalence from programs that do and do not include cases from terminated pregnancies in their data.

Are Comparable Measures Used to Summarize Data?

Once it is decided what to count and how to collect the data on what is being counted, it is important to ensure that the measures used to present the resulting information are the same. If the presenter is calculating the measures from base data, the same measure (e.g., birth prevalence expressed as cases per 10,000 live births) should be used for each of the different population groups, areas, or time periods. However, if the presenter is compiling or comparing already calculated measures, it is prudent to understand how these were calculated. For example, several surveillance programs within the NBDPN have presented birth prevalence as cases per 1,000 live births, while others have used cases per 10,000 live births. This difference should be quite evident in most instances. Less evident is the fact that surveillance programs in the NBDPN tend to use only live births in the denominator (see Chapter 8 Statistical Methods), whereas reports from other groups, such as the International Clearinghouse and EUROCAT, may include spontaneous fetal deaths and/or pregnancy terminations in the denominator. While the inclusion of these outcomes in the denominator will not have the same impact as if they are included in the numerator, it will result in slightly lower prevalence values (Sever, 2006).

When comparing groups within a population it is also good to ensure that specific birth prevalence is being calculated, i.e., that both the numerator and denominator are restricted to the same population. Occasionally, we find prevalence figures where the denominator is based on the whole population and the numerator comes from a subgroup. The above issues can be checked by carefully reviewing the Methods section of the reports from which data are being drawn.

11.3.4 Approaches to Measuring Occurrence—Prevalence Versus Incidence

Birth defects arise developmentally within the first few weeks after conception. As a result, many affected embryos (i.e., cases) will spontaneously abort before a woman is aware she is pregnant. Consequently, in epidemiologic terms, it is impossible for one to reliably assess the *population at risk*, as the number of pregnancies that reach the critical gestational phase where a given birth defect can arise is unknown. In addition, it is unknown how many of these affected pregnancies result in spontaneous abortions. As discussed elsewhere, it is not possible to accurately estimate the *incidence* of a birth defect—the number of new cases of a defect occurring in a population at risk during a specific time period—because one

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cannot establish the number of new cases of the birth defect nor the population of conceptuses that were viable (and thus "at risk") at the relevant point of development (Mason et al., 2005). Most epidemiologists in the field suggest that data be presented and discussed in terms of prevalence, often reported as *prevalence at birth* or *birth prevalence*.

As noted, in reporting the occurrence of birth defects, prevalence estimates are often calculated so that the numerator includes cases that do not appear in the denominator. For example, while the denominator commonly consists of the number of live births, if data are available, it is generally preferable to include birth defects observed among fetal deaths and induced terminations in the numerator. The resulting prevalence is a ratio, which generally includes a multiplier—typically 10,000—so that the reported prevalence of most defects will have at least one unit to the left of the decimal point. Numerically 1.6/10,000 is the equivalent of 0.16/1,000. For further detail see Chapter 8 "Statistical Methods."

Birth prevalence provides a method of expressing the occurrence in a population in a way that supports comparisons. When the number of live births is used as the denominator, to be meaningful it should represent the same geographic and temporal "population at risk" that the birth defects cases come from. For example, in Missouri in 1989–1995 there were 193 cases of tetralogy of Fallot delivered statewide. This provides the numerator for the calculation of prevalence. The 532,592 live births delivered statewide in 1989–1995 are the denominator. The number of cases (193) divided by the number of live births (532,592) times the multiplier of 10,000 yields a prevalence of 3.62 case per 10,000 live births.

11.3.5 Level of Focus

Different types of birth defects can have different causes and arise through several different biologic pathways. Moreover, an individual child can have defects in multiple organ systems. This creates another fundamental issue, which can be thought of as the choice of level of focus: whether the focus is on individual (specific) birth defects or on individuals with birth defects. When the focus is on *individual birth defects*, the occurrence of specific birth defects is of interest. In contrast, when the focus is on *individuals with birth defects*, one is interested in the issue of how many people have birth defects.

How one chooses between these approaches depends on the question being asked or how the data will be used. If one is interested in identifying possible teratological effects of environmental contaminants, for example, the focus may shift from a single birth defect to the occurrence of any potentially related birth defect. This may involve examining the potential association between various chemicals and the occurrence of all types of birth defects.

Many scientists argue that analyzing all birth defects together rather than examining specific defects is of limited value. Importantly, how different types of defects can be aggregated in a biologically meaningful way is an issue of interest. Just as reports on infectious disease do not look at infectious diseases as a group, but present information on specific diseases (measles, shigella, AIDS, syphilis, toxoplasmosis, malaria, etc.), birth defects should be examined in the same way. For example, the epidemiology and causes of outcomes such as neural tube defects, gastroschisis, and Down syndrome are different so the logic of lumping them together may be questionable. Nevertheless, approaches for grouping defects in biologically and etiologically meaningful ways are being pursued.

In addition, it is important to recognize that many times a child will have more than one type of birth defect. For example, 58% of children in the Texas registry have more than one birth defect. Therefore, reporting the numbers of cases of individual types of birth defects, without informing the audience of the extent of multiple diagnoses, may unintentionally lead to an overestimation of the number of individuals in the population with birth defects. Furthermore, many audiences may be specifically interested in the number of persons with birth defects, since this information can be relevant for advocacy and health planning purposes.

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11.3.6 Risk Factors and the Importance of Timing with Respect to Exposures

Surveillance programs often collect limited data on risk factors for birth defects, as well as on cases. However, there are important distinctions between those types of data routinely collected and those obtained as part of special studies (such as cluster investigations) or in conducting epidemiologic research. This discussion focuses on risk factor data that are often collected routinely and their presentation.

Exposures known to be risk factors for birth defects are quite limited, one of the issues that makes additional epidemiologic studies so important. Examples include *maternal metabolic imbalances* (such as diabetes) and *viral infections* (such as rubella), as well as a small number of *drugs* and *occupational/environmental chemicals*.

Three sociodemographic variables for which data are routinely available can potentially be considered risk factors for some birth defects. These are maternal age (date of birth), maternal race and ethnicity, and maternal education. As discussed in Chapter 4 (Data Variables), the first two of these are considered as core variables, while the last is a recommended variable.

In presenting data on these variables, *maternal age* is usually grouped either into quinquennia (\leq 19, 20–24, 25–29, 30–34 and \geq 35) or into two age groups (<35 and \geq 35). The latter grouping is used as it is particularly relevant to Down syndrome risk and prevalence. As discussed in Chapter 4, *race and ethnicity* should be presented in categories that are comparable with the federal standards in current use. If data on *maternal level of education* are collected, then they should be presented in the same categories used on the birth certificate. Following these recommendations in presenting data on the above sociodemographic variables allows information on cases to be compared with that from the certificates of live births for the at-risk population.

A final type of risk factor information to be considered here is *maternal place of residence* (address). This, too, is considered a core variable and is basic to the use of geographical information systems, a topic discussed later in this chapter. In terms of presenting data on residence, cases are usually aggregated into some geopolitical unit (such as counties) or into administrative units (such as health regions), for which information on live births is available. How these aggregated data are presented to the public or to data users other than surveillance program staff is considered below in the discussion of data suppression.

While perhaps not pertinent to the way data are presented in a general sense, the issue of maternal residence as a risk factor raises an important point about presenting information in epidemiologic studies. Particularly with the increasing utilization of GIS, the location of the mother's place of residence is sometimes used as a surrogate for exposures in studies of risk factors associated with the ambient environment (Sever, 1997). In considering residence as a surrogate for exposure in studying birth defect risk factors, it is important to know the location of the mother's residence at the time in gestation when relevant developmental events are occurring. Periods of sensitivity are well known for many organs and structures and, for the most part, these are during the embryonic period, early in pregnancy (Mortensen et al., 1991).

Unfortunately, most surveillance programs collect only information on the mother's address at the time of delivery, when it is residence during embryogenesis that is biologically relevant. This is important in assessing possible risks related to the ambient environment because several studies have shown that a

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large percentage of women move between conception and delivery (Canfield et al., 2006). Residence at delivery, therefore, is not only limited in its usefulness as a surrogate for exposure, but in many cases it does not reflect biologically relevant exposure, since it does not represent where the woman lived when crucial events in embryogenesis were taking place. This limitation should be noted when data on maternal residence are presented as part of epidemiologic studies of environmental reproductive hazards.

11.3.7 Privacy and Data Suppression

Specific birth defects are often rare events (sometimes extremely rare) leading to yet another set of issues that must be considered when presenting birth defects data. The public health professional must balance the potentially conflicting goals of information dissemination with protection of the privacy of persons in the community. When the number of cases in a diagnostic category within a group or stratum (such as race or sex) is small or the population from which the cases are determined is small, the risk of allowing a specific individual to be identified may be deemed too large to be acceptable. In such cases, steps must be taken to protect an individual's privacy. In addition to protecting privacy, prevalence information is often suppressed when concerns exist regarding possible statistical unreliability of estimates that are based on small numbers.

The most common method of preventing the identification of specific individuals in tabular data is through *cell suppression*. This means not providing counts in individual cells where doing so would potentially allow identification of a specific person. Cell suppression can also be done by combining cells from different small groups to create larger groupings that reduce the risk of identifying individuals. While there are also more sophisticated data perturbation methods that use statistical noise to mask sensitive information, these are generally more suitable for use with economic or financial data than with public health data.

In general, the more restrictive a suppression rule, the less information a given table or report will provide. The weaker a suppression rule, the greater the potential threat of revealing confidential health information. It is a question of balancing the threat to individual privacy with the public health value of presenting the data.

Overall, deciding when and how to suppress birth defects information is more a social, political, and legal issue than a technical one. The technical aspects are quite straightforward, but the contextual and procedural/policy issues are likely not to be. These all need to be considered and balanced in the local context before informed decisions can be made to suppress or not to suppress data in program reports or other documents.

Surveillance program administrators and technical staff should be aware that standards used to suppress data may already be set in state laws or in departmental or institutional rules and regulations. It is the responsibility of surveillance staff and administrators to be aware of these standards and practice within their limits. If standards are not established, it behooves a surveillance program to establish rules that will be followed consistently. This is best accomplished with the assistance of an advisory committee, an institutional review or privacy board, or a similar body.

Appendix 11.1 reviews the basic methods, issues, strengths, and vulnerabilities of cell suppression.

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11.3.8 Geographic Information Systems (GIS)

The application of Geographic Information Systems (GIS) methods has become an integral component of aggregating, analyzing, evaluating, and displaying health data. The current practical applications of GIS in epidemiologic studies range from descriptive statistics (i.e., plotting data on a map) to evaluation of spatial relations between environmental exposures and health outcomes.

Several definitions exist for geographic information systems. One of the most recent, as found in *Healthy People 2010*, defines GIS as "powerful tools combining geography, data and computer mapping" (U.S. Department of Health and Human Services, 2000). Software packages available today, such as ArcMap and MapInfo, integrate many GIS functions. These include (1) database management, (2) data manipulation and analysis, and (3) data presentation (i.e., displaying data on a map). To be included in GIS, the data should have some kind of geographical or spatial component that can be translated into digit maps. Appendix 11.2 contains a brief introduction to GIS mapping along with a list of suggested references.

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11.4 Stage 3 – From Information to Knowledge

ACTORS NATURE OF PRESENTATION PRODUCT **PRODUCT TYPES** MODE **Knowledge that is:** Knowledge Knowledge Makers: **Types** Generated through iterative, multi-Data providers directional Presentations Discussion communication tailored to Data interpreters information needs between Written feedback Action takers collectors, and backgrounds on presentations of various interpreters, and audience types users of data

As mentioned previously, the key themes of the CDC definition of *surveillance* are the integration of data collection, analysis, interpretation, dissemination, and application. In the previous section we spoke of moving from analysis to interpretation, whereby data are converted to information. In this section we are more concerned with dissemination of information with an eye toward application, whereby information is converted to knowledge capable of informing action. We now turn to some of the more technical, as opposed to the more philosophical or theoretical aspects of data presentation. In the broadest sense, we are here concerned with the clarity of the information presented and a lack of ambiguity in the message to be communicated.

We can conceive of the process of communication as having five major components—the sender (presenter), the medium, the message, the objective(s), and the receiver (audience). It is important in the development of a data presentation to keep all of these components in mind. It is also important to realize that communication is not simply a linear process of conveying the message from the sender to the receiver, but rather often involves a loop from the sender to the receiver, back to the sender, and back to the receiver.

In general, we suggest working backward through the communication sequence when designing your presentation. That is, instead of beginning with yourself (the sender) and what you want to tell the audience, begin by thinking about the audience (the receiver) and its information needs. Beginning with the audience will help you determine the objectives of the presentation, formulate the message, and select the best medium to use in conveying that message. Below we walk you through the process of developing a data presentation by (a) accurately characterizing the audience and understanding its needs, (b) establishing the purpose or objectives for a given presentation, (c) developing the content of and ensuring the clarity of the message, (d) selecting the most appropriate medium for the message, and (e) being aware of biases you as the presenter may have. We do not mean to suggest that consideration of elements a-e must be undertaken sequentially. However, all need to be considered carefully in the context of the presentation as a whole, even if some are apparent "givens". For example, if you are told you must prepare a report for the Governor on x topic, then you know the audience and the medium as well as the overall objective of the report, namely "to provide information on x." Still you would do well to learn

more about why the Governor is interested in x, what specific information is being sought, and how the information will be used before developing the report.

11.4.1 The Receiver—Understanding the Audience and Its Information Needs

Know your audience! One of the central tenets of any presentation is identifying the audience being addressed and recognizing the information needs of its members. This includes taking into consideration the audience members' backgrounds, interests, and bases of knowledge. For example, a presentation to epidemiologists may include detailed information on complex analyses, yet these should be presented only as a summary to an audience of policy makers. The former may expect—and insist on—a presentation including numerical estimates of standard errors, confidence intervals, etc., while the latter will respond better to straightforward graphical displays that illustrate the key points. Even in an apparently homogeneous audience there can be significant heterogeneity. For example, a presentation to a parent group may include both highly informed individuals who have extensively researched a particular birth defect, as well as new parents who may be wholly unfamiliar with the field.

If the nature and level of expertise of your audience is not clear to you, do not hesitate to talk to someone in a position to know more about the audience and why the presentation has been requested or arranged.

11.4.2 The Objective(s)—Determining the Purpose of the Presentation

The type of information an audience is interested in and the questions posed can vary considerably, which in turn will influence your objectives in developing the presentation. An audience consisting of policy makers may be hoping to learn about population trends and attributable risk. Researchers may be interested in the prevalence of cases based on various demographic variables, while service providers may be most interested in the geographic distribution of cases and services. These differences lead to different types of questions that will require different analytic approaches and may lend themselves to different formats of data presentation. In Appendix 11.3 (the Data Users Matrix) we characterize a number of possible audiences for a birth defects surveillance presentation in terms of their likely information needs and presentation approaches that might meet those needs.

In sum, one must be prepared to use different approaches to audiences that differ in current levels of knowledge regarding the topic, as well as in having different interests, objectives, and information needs. The questions of interest to a particular audience will drive both the analytic approaches and the medium or format selected for presentation.

11.4.3 The Message—Developing Content and Ensuring Clarity

Having meticulously collected, cleaned, and analyzed a surveillance program's birth defects data, the proud owner of neatly tabulated findings may well wonder, why it is necessary to also express these findings in graph or chart form. Shouldn't the numbers speak for themselves?

The answer is yes, of course, the researcher should be able to verbally convey the most important results and to summarize succinctly characteristics of the data. In addition, it is certainly helpful to make complete tabular data available to the consumer of epidemiologic results (i.e., the audience). However, while individual learning styles differ, most people are primarily oriented to interpreting visual information as opposed to tabular data (Spence, 1990) and can more easily make judgments about that information based on a limited number of simple cues: smaller/larger, brighter/darker, increasing/diminishing. Therefore, a graphical display increases the efficiency with which your audience processes your information (Legge et al., 1989). Remember, too, that data presentation is aimed at

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meeting a specific purpose; whether stated or not, you have an objective and a message to convey, and your audience needs to understand it.

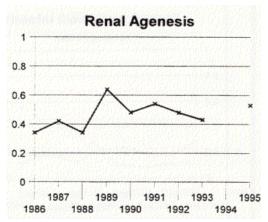
In this section we discuss graphical representations (such as graphs and charts), concluding with tips for you to keep in mind as you develop a data presentation. We then offer guidance on how to choose the appropriate format for displaying a given type of data, with further detail provided in Appendix 11.4. We conclude this section with a discussion of the characteristics of a clear, informative table.

Graphs and Charts

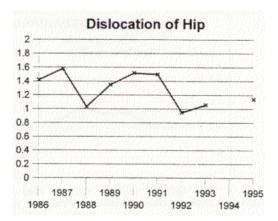
The discussion below will enable you to create graphical representations of your data that meet the following requirements:

- > Convey results accurately
- ➤ Allow for efficient interpretation
- > Engage the interest of the audience

Conveying results accurately. Essentially, all of the information conveyed through graphs and charts allows for comparison and answers a single question: which is larger? This is a question of proportionality. Therefore, it is important that visual elements reflect the same proportions as the data they represent. For example, Sample Figures 1A and 1B demonstrate cases per 10,000 live births for a specific birth defect, but the figures use a different range of values on the y-axis. This practice distorts the actual differences in proportion making it appear as if the rates of these two defects are quite similar, when in fact dislocation of the hip is about twice as common in this population (Muscatello et al., 2006).



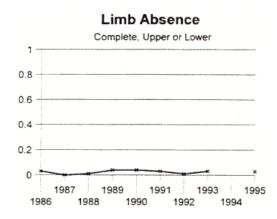
Sample Figure 1A. Cases per 10,000 live births 1986–1995



Sample Figure 1B. Cases per 10,000 live births 1986–1995

However, it is not always desirable to use the same scale for all charts. Sample Figure 1C demonstrates cases per 10,000 live births using the same scale as Sample Figure 1A, but since absence of limbs is so much rarer than renal agenesis, it is difficult to detect any difference among years for Limb Absence. Therefore, it is important to weigh the essential information you want to convey before deciding on scale (as well as other features); in this case, which is of primary concern: between-defect comparisons or illustrating a trend for one particular defect?

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Sample Figure 1C. Cases per 10,000 live births 1986–1995

Chart design characteristics that can distort proportions when changing scales across multiple graphs include:

- ➤ Two different graphs examining the same outcome, but based on different time periods or different lengths of time.
- > A bar graph of several time-based groups, where the groups correspond to different lengths of time
- ➤ Graphs of statistical functions, such as regression lines, that extend beyond the range of values observed in the data.
- > Use of three-dimensional graphical elements.

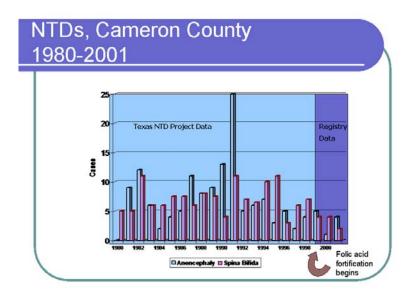
Allowing for efficient interpretation. To support efficient interpretation of data an important principle to follow is the *ink-to-data ratio*. Simply put, try to minimize the proportion of "ink" (or what would be ink on a printed page) that is employed in actually representing data. This means eliminating extraneous graphical elements that do not convey additional meaning, such as slide backgrounds, clip art, animations, and other elements of what is often referred to as "chart junk."

Chart junk can appear in two varieties. The first is extraneous material unrelated to the actual data. This type of junk is relatively easy to eliminate as it tends to be under the control of the person using the graphing software. So resist the temptation! In cases where the junk is generated by the graphing software, do not hesitate to edit it out wherever possible.

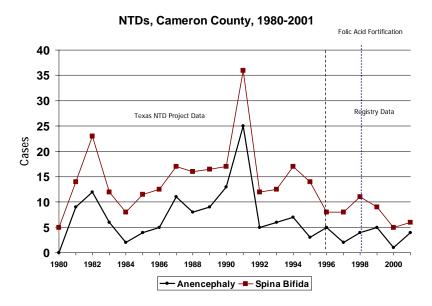
The second form of chart junk involves certain graphic styles that require a large amount of space to convey a small amount of data. In this regard, the key is to focus on the data themselves, rather than the data "containers." Data containers are shapes used to reflect data, such as bars and line markers, and minimizing their size can be particularly helpful if one is presenting a large volume of data.

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For example, consider which of the figures below is easier to understand, Sample Figure 2A or 2B. *Hint:* See how many instances of chart junk you can identify in Sample Figure 2A.



Sample Figure 2A. Example of excessive "ink:data" ratio

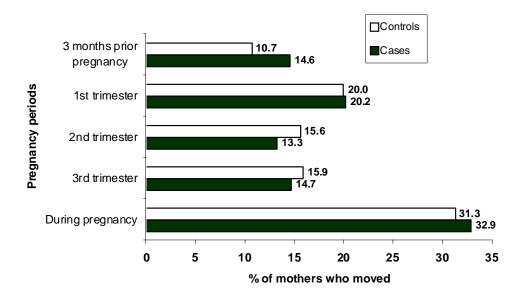


Sample Figure 2B. All "ink" conveys essential information

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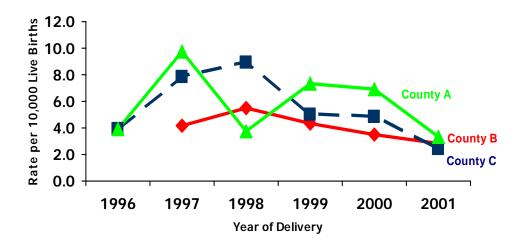
¹ In Sample Figure 2A, the differing color backgrounds, the slide design elements, and the stylized arrow, combined with 3D bars, employ a great deal of "ink" to convey the same information as conveyed in Sample Figure 2B.

Sample Figures 2C through 2F present further techniques to reduce the amount of "ink" in a chart or graph. In 2C, adding data labels to the bars allows you to eliminate additional "ink" in the form of gridlines, while allowing the viewer to accurately assess the value of each bar. Horizontal orientation allows category labels to be spelled out rather than abbreviated. Sample Figure 2D contains no legend; rather each data series is labeled directly, with color coding used to ensure correct pairing of label with series. Sample Figure 2F (versus 2E) also uses direct labeling instead of a legend, and changes X axis scaling to every other year, which is sufficient for these data.



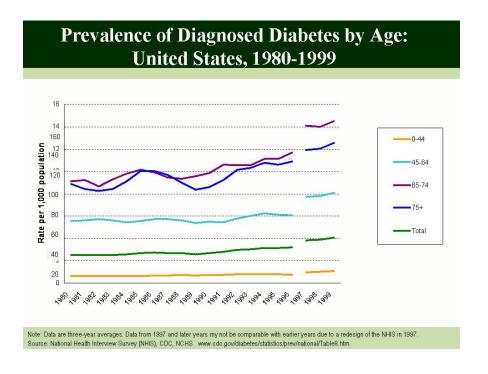
Sample Figure 2C. Use of data labels to eliminate additional "ink" in the form of gridlines. Horizontal orientation allows category labels to be spelled out.

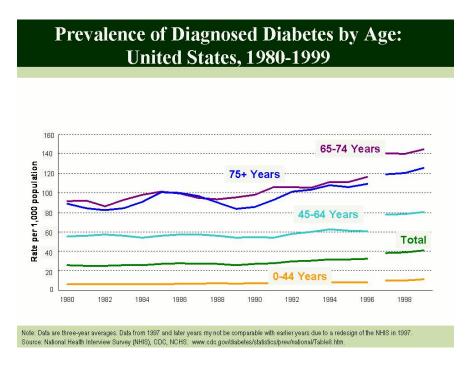
NTDs by County, 1998-2001



Sample Figure 2D. Direct labeling of data series rather than legend. Use of color coding to ensure correct pairing of label with series.

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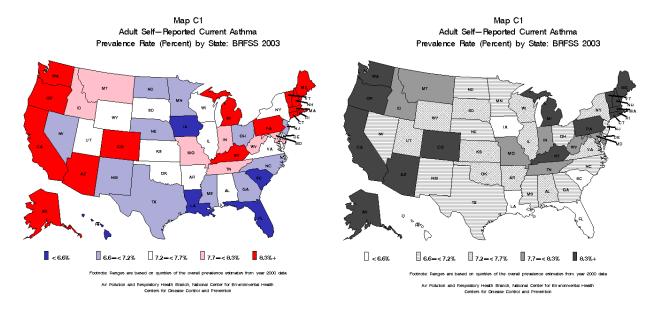


Sample Figures 2E and 2F. Note that Figure 2F uses direct labeling rather than a legend, and changes X-axis scaling to every other year.

As a general rule, an intelligent reader/observer should be able to clearly interpret a chart or graph without referring to supplemental text or materials. If a figure will be used in a live presentation, the information presented visually can be minimized to the extent that it will be supplemented orally. However, copies of an oral presentation or figures used in formats without benefit of augmentation by a presenter should contain sufficient information to stand alone yet still be understood.

Engaging audience interest. While tabular data lend themselves to accurate interpretation, especially by those accustomed to working with numbers, they nevertheless require more time to process (Spence, 1990), are tedious to follow in a slide presentation, and are less accessible to non-technical audiences. A compromise suggested by Tufte (2003) is to use handouts, including the actual data tables, in lieu of the standard 2x3 printed version of slides.

Cautions about chart-junk notwithstanding, certain visual elements can improve audience engagement. For example, color can be an effective means of increasing visual interest and adding clarity to a figure (compare the differing impact of Sample Figures 3A and 3B). Color can also be used to portray increasing data density (the amount of information conveyed relative to the size of a figure) or to add an additional level of information to a figure. For example, the size of a dot may indicate the number of babies born at a hospital, while the color of the dot indicates the percentage of births who spend more than 24 hours in a neonatal intensive care unit. However, avoid too much color, as well as combinations of colors that may distract, confuse, or mislead readers.



Sample Figure 3A. Example of a map using color codes

Sample Figure 3B. Example of same map in gray scale

Understated, subtle backgrounds, textures, and other graphical elements can be eye-catching but can also easily be over-used. Furthermore, no amount of visually stimulating material on a chart can take the place of a presenter whose tone of voice, bearing, and engagement with the audience bespeak a clear understanding of and excitement about the information being presented. Table 11.1 below contains some summary tips for graphical data presentation.

Table 11.1 Summary Tips for Graphical Data Presentation

General Tips

- Remember that the default graphing settings on your software package (e.g., PowerPoint) are rarely the best for creating an effective graph. If you do not have the time or interest to customize your own slides, consult an expert in your organization.
- Use a clear and simple font (e.g., a sans serif font such as Arial).
- Use footnotes to explain acronyms and methods (Muscatello et al., 2006).
- Restrict the use of abbreviations to those that will be known to everyone in a potential audience or readership, or provide a list of the less well-known abbreviations used, keeping them few in number and usage.
- Indicate the units that are being used (e.g., age in days, weight in grams).

Analytical Tips

- Emphasize differences between groups—identical patterns across groups can be stated and/or expressed in a bullet point and do not need to be portrayed in a figure.
- Avoid comparisons across multiple figures.

Visual Tips

- Avoid the use of background pictures, or additional pictures, lines, or shapes that are added solely to "beautify" a figure.
- Avoid the use of unnecessary or heavy gridlines. Use white spaces with a bar instead of a grid line.
- Eliminate 3-D bar graphs, which add lines and shading while providing no additional information. Furthermore, two-dimensional charts are generally interpreted more quickly and accurately than those in 3-D (Hughes 2001).
- Eliminate unnecessary legends. Legends—if absolutely needed—can be placed inside
 the plot area for a graph. This increases the maximum size of the graph. Rather than a
 legend, use direct labeling if possible.
- Simplify labeling (Muscatello et al., 2006). For example, a time series on the X axis need not always have every year listed—it is implied that 1995 is the point between 1994 and 1996.

Staying on Message

- Remember your core message and do not present irrelevant data (e.g., detailed methodological information if not a methodological study).
- For certain audiences (e.g., lay persons or policy makers), consider wording the title as a plainly stated question that guides interpretation of the graph (Muscatello et al., 2006). For example, "Is gastroschisis more common among babies born to younger mothers?" rather than "Patterns of prevalence of gastroschisis by age of mother"
- Show your charts and tables to someone unfamiliar with the data and ask them how they interpret the "bottom line" message from each. Revise to improve clarity.

What Type of Graph or Chart Should I Use?

Appendix 11.4 contains information on some of the more common types of graphs and charts along with suggestions on how to choose a type appropriate to the data you are planning to display.

Before making your final decision, however, you should also ask yourself two questions that relate less to the nature of your data and more to your own personal preferences and the needs/interests of your audience:

- Am I comfortable explaining this graph or chart? If the answer is no, find an alternative format with which you are more comfortable.
- ➤ Given my audience, should I sacrifice detail for clarity, or clarity for detail? For example, an audience of foster parents would probably benefit from clarity with less detail, whereas an audience of epidemiologists will readily comprehend your meaning and will rather be looking for additional detail about methods or sample characteristics.

Tables

Despite the usefulness of graphical data presentation formats such as those just described, there will be times when a table is still the ideal choice. Tables display data in a systematic way and help readers locate specific information readily. Simple tables can stand alone in a slide presentation or be used as a supplemental handout when presenting summary data in graphical format.

Good tables have (see Sample Table 1):

- ➤ A table number
- A table title that clearly identifies the data displayed
- Column and row headings
- At least 3 horizontal lines (below the title, column headings, and data fields)
- Decimal alignment
- Expanded forms of abbreviations used in the tables, generally as footnotes
- ➤ Additional explanatory footnotes as needed

Sample Table 1. Counts of selected birth defects cases and maternal country of birth, 2004

	Maternal country of birth							
	U.Sborn*		Mexico/CA**		Others***		Missing	
	count	%	count	%	count	%	count	%
Controls	539	48.4	498	44.7	68	6.2	8	0.7
Heterotaxia	63	36.6	97	56.4	11	6.4	1	0.6
Omphalocele	42	48.3	44	50.6	0	0.0	1	1.1
Gastroschisis	58	43.3	63	47.0	12	9.0	1	0.7
Oral clefts	49	52.1	38	40.5	7	7.4	0	0

CA=Central America

- * 50 U.S. States, Puerto Rico, Virgin Islands (U.S.)
- ** Mexico, Costa Rica, El Salvador, Guatemala, Honduras, Nicaragua, Panama
- *** Algeria, Argentina, Bolivia, Brazil, Others

11.4.4 The Medium—Ensuring Its Appropriateness

Now that you have considered your audience and its needs, established the purpose for communicating your data, and developed the content and clarity of the message, it is necessary to select the most appropriate medium for the message so that it reaches the right people in a way that will help them to understand, interpret, and use the information. The selection of an appropriate medium, or communication channel, varies depending on the format of your message and the audience's access to the medium.

Communication channels can be active or passive. *Active channels* require the audience to engage with the information; *passive channels* require less interaction. Interpersonal communication, print readership, and Internet communication are examples of active channels, while passive channels include television and radio. A study comparing media type and source of information with the personal context of health-oriented attitudes and behaviors (Dutta, 2007) has demonstrated that health-oriented individuals sought active channels as primary sources of information. Non health-oriented individuals were more likely to obtain information, such as prevention messages, through passive entertainment-education channels.

Information might reach your intended audience directly, via publications, or more indirectly, such as through interpersonal communication by a social service professional relaying information to a family affected by birth defects. As you communicate information through one channel, consider how the data will be interpreted as they flow through other channels (Valente et al., 1996). Below we briefly discuss some of the more common communication channels used for the presentation of birth defects data.

- Reports and publications
- Professional presentations
- Mass media
- Websites
- > Community outreach

Reports and Publications

Birth defects data are commonly presented in reports, including internal documents, working papers, and scientific publications. Use guidelines from journals for content and format. The level of detail should be based on the audience and its needs. Follow the principle of *tell 'em:* "Tell 'em what you're going to tell 'em, tell 'em, and tell 'em what you told 'em" (Collins, 2004a). Summarize the key points of the report in an abstract or executive summary, highlight your message clearly, and conclude with a summary. A well-written abstract should be able to stand alone without reference to the article or report being summarized and should concisely outline all relevant topics while excluding unnecessary detail, generally in 200 words or less.

Within the report, pay careful attention to describing explanatory table headings and figure legends. A review of graphical presentations published in *Journal of American Medicine* and *Annals of Emergency Medicine* (Cooper et al., 2002) identified few indicators of poor quality graphs: lack of definition of symbols, internal errors, contradictions with the text, numeric distortion, lack of visual clarity, nonstandard graphic conventions, or extraneous decoration. However, 31% of graphs were not self-explanatory, meaning the reviewers could not unambiguously interpret the graph despite reading the study design and legend of the graph. Additionally, 48% of graphs did not illustrate the underlying distribution and 48% did not depict important covariates.

Professional Presentations

At professional meetings, data are generally presented as poster presentations or platform presentations.

Poster presentations. A poster presentation is a visual display that summarizes your research or programmatic project. The display is mounted on a poster board provided at the meeting. The display includes visual aids such as data tables, charts and photos, along with a limited amount of text presenting the highlights of your topic. Conference participants should be able to quickly understand the work you are presenting including, as appropriate, your central research question or hypothesis, your research approach, and your results. After reviewing your poster, many participants will ask you questions and share their observations. Poster presentations can be an ideal way to:

- Provide a limited amount of information to a diverse audience
- > Start productive conversations with new colleagues
- > Summarize work you have recently completed
- > Obtain useful feedback in developing the study further or in developing a manuscript
- ➤ Advertise your work to colleagues or potential employers

Poster presentations provide key opportunities for scientists to network and discuss shared interests with colleagues.

Successful posters tell an interesting story and are visually appealing, logically organized, and easy to read. Visually appealing posters are simple, uncluttered displays that use a variety of tools to convey information (e.g., data tables, figures, photographs). Color adds interest, but be conservative about the number of colors you use. Bright colors can be disconcerting. Judicious use of underlines, boldface type, and bullets can succinctly highlight important information. "White space" is critical to creating an uncluttered look. A poster printed on a single large (8' x 4' or 4' x 4') sheet of paper is the easiest to view and mount on the poster board.

When constructing figures, charts, or tables, focus the viewer's attention on the data by reducing or eliminating chart "junk" such as non-essential lines or redundant percent symbols (%). Limit the number of decimal points presented. When you can, label data directly rather than referring the reader to a legend. If possible, convert tabular material to figures that are easy to understand. (See Section 11.4.3 for further discussion of charts and graphs.)

Logically organized posters start with a banner title across the top with the authors listed below, followed by their institutional affiliations. Poster content—text and visuals—should be organized so that they begin in the upper left corner of the poster and end in the bottom right corner. Readers will look at the poster from the top down and from left to right. The layout should follow the format of your conference abstract: generally covering the topics introduction or background, methods, findings, and conclusions. Many posters include the abstract as the initial block of text. Each section should have a brief heading, and sections should be separated by a little "white space." The text should be condensed to key points and grouped into blocks of no more than 50–75 words. Avoid abbreviations or acronyms that may be unfamiliar to your viewers.

Posters that are easy to read use fonts that are legible from a distance of 3–5 feet. For the poster title, use a very large font (84 point or larger). Author name and affiliation information can be displayed in 72 point.

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For other elements of the poster, consider these guidelines:

- ➤ Headings and subheadings at least 32 point
- > Text, figure legends, and tables at least 18 point

Keep the font style for similar content consistent throughout. Be sure that format headings and text of the same level of importance use the same font size. Avoid upper-case or "ALL CAP" fonts. Dark letters on a light background are easiest to read.

Some people may ask that you "walk" them through your poster. Avoid reading it! Instead, summarize the big picture of what you did and why. Use the poster's graphics to illustrate your major findings and support your conclusions. Presenters often provide a condensed version of their poster for interested viewers (e.g., a PowerPoint handout). You might also consider handing out additional information, such as supplemental data tables. *Always include your contact information*.

Platform presentations. Platform presentations are delivered through a structured talk or lecture, commonly using presentation visual aids, such as MS PowerPoint. Effective PowerPoint presentations *support*, rather than replace, the delivery of your presentation. Do not be tempted to read directly from your slides. The quality of the presentation depends on the quality of the presenter's communication of the information and not entirely on the quality of the visual aids (Collins, 2004a).

As with any public speaking activity, speaking softly, unclearly, or in a monotone voice; using excessive hand gestures; and speeding through slides without giving the audience a chance to digest the information will not communicate your message well. Pay attention to the pace and timing of your talk, allowing pauses but also following time limits. Prepare your presentation for compatibility with any computer, bring back-up copies of your presentation and, most importantly, rehearse. Rehearsing, especially in front of a representative audience, will help you become comfortable with your presentation, provide an opportunity to clarify any points that are potentially confusing, and enable you to assess the presentation's natural and logical flow (Collins, 2004b). It will also give you another chance to proofread for potentially embarrassing errors.

When preparing your visual aids, follow principles of clarity, readability, and simplicity. For clarity, design your slides with only a few key points per slide. A standard recommendation is the "rule of six": 6 lines per slide and 6 words per line (Collins, 2004b). Use contrasting background and text colors so your words are readable, but avoid hard-to-read color combinations such as red/green, brown/green, blue/black. Font sizes should be at least 24 pt for text and 36–40 pt for titles, but also consider the size of the room you are presenting in to ensure the people furthest from the screen can read the slide. Setting the entire text in bold can also increase readability.

In terms of simplicity, emphasize the most critical point on each slide. Include pictures and graphs for visual interest when they are relevant, but choose them wisely to minimize distraction from the main point. Tables can be difficult for audiences to read and interpret; look for other ways such as graphs or text to communicate the same information more clearly. If you do choose to use a table, be sure to make use of white space so that the audience can easily see the most salient points without sifting through clutter (Ryder, 1995).

Finally, remember that it is not the topic or data alone that creates a meaningful presentation. Strategic communication of understandable information is the key to successful delivery of data through the professional presentation medium (Thompson et al., 1987).

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Mass Media

Dissemination of birth defects data to the general public occurs through many channels: printed news material, television, radio, and websites are just a few examples. Since these media have a broader reach than presentations at professional conferences, the audiences will be more heterogeneous. It is important to integrate the target audience's cultural values into the strategy when selecting the appropriate communication channel, but the ethical challenges of communicating information accurately through mass media are difficult to avoid (Guttman, 1996). A review of 10 years of health content in the media concludes that "popular media is not likely to facilitate understandings helpful to individuals coping with health challenges" (Kline, 2006). The topic of birth defects tends to be misrepresented in the media, generating unnecessary public anxiety (Marks, 1993). If mass media is chosen as a communication medium, think about how the public understands and interprets risk, so that it is not interpreted inaccurately (McComas, 2006).

While there is no method that will match all needs for knowledge, understanding the needs of potential users will help determine if mass media channels are appropriate as well as the best way to tailor the message through the medium (Williamson, 2005). Communication strategies should consider the audience's access to information channels, motivation for information, literacy and numeracy, likelihood of interpreting complex data, and cultural context.

Websites

Using websites to convey information about birth defects to the public is becoming increasingly common as health-oriented individuals actively seek knowledge, but these individuals' trust in the information source is paramount. Analysis of data from the Health Information National Trends Survey (Rains, 2007) shows that "trust in information-oriented media, entertainment-oriented media, and one's health care provider all predicted Web behavior and perceptions." Users of the Internet as a source of information are most likely to be women who have high knowledge about resources, regardless of format, and are likely to discuss the information they find with health care providers (Warner and Procaccino, 2007). These women typically have a higher level of education and socioeconomic status (Pandey et al., 2003).

Websites are also useful for disseminating data to research, surveillance, program, and policy users. For all audiences, the website should be clearly laid out, interactive, tailored to the audience, and regularly maintained and updated for current information.

Community Outreach

Another way to communicate birth defects data to the public is through community outreach. Think about creative ways of disseminating information in addition to more traditional routes; look beyond PowerPoint, posters, and reports. Your audience could be someone affected by a birth defect who may or may not attend conferences, read journals, or look at websites. As mentioned earlier, non-health oriented individuals may not actively seek information, especially if they have low literacy or numeracy skills, and consequently low health literacy skills. The attributes of health literacy are "reading and numeracy skills, comprehension, the capacity to use information in health care decision-making, and successful functioning as a healthcare consumer" (Speros, 2005). Over 50% of Americans have limited literacy and numeracy abilities according to a 1992 National Literacy Survey so health materials should be written in simple terms to increase understandability. The health literacy approach is not "dumbing down" data, but simplifying it into reader-friendly plain language so the message is communicated clearly (Stableford and Mettger, 2007).

Some examples of community outreach strategies include:

- > Strategically disseminating materials (brochures, posters and pamphlets) in public locations,
- > Delivering information at community events or health fairs,
- ➤ Connecting with key community gatekeepers such as health promoters who share birth defects information through interpersonal communication.

Understanding the local context is imperative for developing appropriate communication strategies for community outreach.

Remember: "A word of caution that can't be repeated often enough: The medium does NOT replace the message, be it Morse code or interactive video-on-demand. The principal objective remains to choose the right message, for the right people, at the right time and to ensure that it gets through in the most efficient and effective manner" (Chamberlain, 1996).

11.4.5 The Sender—Being Aware of Biases

Finally, as a presenter, one rarely faces an audience without having one's own personal interests and objectives. These may range from seeking funding to promoting a particular theory or model and may or may not align with the objectives and interests of the audience. We should nevertheless strive to present information in as impartial and balanced a manner as possible. This includes not omitting or minimizing contrary information, or choosing or manipulating figures or statistics in order to support a given objective.

11.4.6 Pulling It All Together

What are the factors that drive data presentation at the stage when you are transforming information into knowledge? As stated previously, when planning a data presentation, it is important that you as presenter, and catalyst in the transformation, pay attention to all the other elements of the communication process. That is, that you (a) understand the audience and its needs, (b) establish the objective(s) for the presentation, (c) determine—based on earlier analysis and interpretation—what the message is and how most clearly to present it, and (d) decide upon the communication medium. That is, the elements listed below must all be suitably "matched" in a data presentation:

- ➤ Audience and its needs
- ➤ Objective(s)
- ➤ Message (information being shared)
- > Communication medium

The three case studies presented below demonstrate how these elements of a presentation must be coordinated and addressed.

Informing the Public about Birth Defect Prevalence

Audience The public

Objective To inform the public about the frequency of a birth defect in

an area, e.g., a state or public health region

The observed prevalence of birth defects during a specified Message

time or trends over time

Communication Medium Tables or graphs that are clearly labeled, with the terms and

> categories defined so that they are intelligible to the intended audience. The medium could be a published report; a press release, with supporting technical documentation; or a document on the surveillance program's website.

Informing Policymakers about Birth Defects Issues

Audience Legislators or policy makers

Objective To support efforts to increase health services or justify

continuation of funding for the surveillance program itself.

Message The magnitude of a problem or the resources needed to

maintain a surveillance program.

Communication Medium Clear, succinct bulleted text with supporting graphs and

tables.

Responding to Community Members about Birth Defects Clusters

Audience Community members

Objective To respond to concerns about birth defects clusters

Message Relationship (if any) between birth defects clusters and

environmental hazards

Communication Medium Established state protocols for dealing with this issue and

> including description of how information regarding the cluster and its investigation is communicated to concerned stakeholders. Important to communicate information to the community, both during the investigation and at its

conclusion, using clear and simple messages (Williams et al.,

2002a).

11.5 Stage 4 – From Knowledge to Action

ACTORS NATURE OF **PRODUCT PRESENTATION PRODUCT TYPES** MODE **Action Takers Actions that** Action Types are: Surveillance reports Data reporting staff Estimating Websites frequencies Surveillance staff Appropriate Scientific Referrals to Policymakers Evidence-based publications services Decisionmakers Maximally Policy papers Planning services Intervention effective and cost-Guidelines designers/ Planning effective implementers interventions Intervention Health care protocols Conducting providers research •Risk Media communication Cluster Families investigations Press releases, Community media articles and members shows •Fellow scientists/ researchers

As mentioned earlier, the key themes of the CDC definition of *surveillance* are the integration of data collection, analysis, interpretation, dissemination, and application. In the previous section we spoke of dissemination of information with an eye toward application, whereby knowledge capable of informing action is developed. In this section, we speak of application or the undertaking of action(s) in order to achieve programmatic objectives. To illustrate this stage, we present a vignette of a surveillance program as it moves through different developmental phases (nascent, developmental, mature) and how the data produced at each phase of a program's development can be mobilized to inform action.

The stage of development of a registry or surveillance program has important implications for data presentation. The following vignette describes the experiences of one program director in this regard. The text is in the first person to reflect the program director's appraisal of the events surrounding the presentation of data to different audiences at different developmental stages of the program and with different types of action likely to result.

In my experience, the quality of our data increased dramatically from our initial data set to the second and has increased incrementally thereafter. We are continuously evaluating our methods and data, with the goal of being more complete, more accurate and reducing bias. Nonetheless, I believe all of our data have had some value and were worth presenting to selected audiences.

I received our first data set the day I was asked to take responsibility for the State of Contentment's birth defects surveillance program. I was handed a flexible folder that in essence

was the registry. In it were a couple floppy disks, several sheets of paper with diagnoses listed on them, and a couple of envelopes containing various parts of copied discharge sheets. Not an ideal data set, but it was the result of a pilot project where hospitals in one region of the state reported their birth defects cases from one year to the department of health. The regional perinatal center had prepared a formatted Excel spreadsheet for the project data, but they were the only hospital to use it. While not standardized and not complete, these were the best data we had at the time.

We compiled the data into a table based on the tables of birth defects in the NBDPN annual report and presented them at a meeting organized by the local chapter of the March of Dimes. The meeting coincided with the March of Dimes' annual legislative lobbying day. It was a relatively informal meeting, and we provided handouts of the data to a mixed audience made up primarily of March of Dimes volunteers; a number of neonatal intensive care unit (NICU) nurses, geneticists, and neonatologists also attended the meeting. The March of Dimes was particularly interested in the data, as they had lobbied the legislature to establish a birth defects surveillance program, legislation which included authorization of the pilot project. The presentation was informal, accompanied by a warning that the data were very messy and likely to be incomplete. Nonetheless, the audience was enthusiastic. The volunteers asked a lot of questions, as did the professionals who also offered a good deal of advice. Among other things, I recall learning the importance of using standardized case definitions; the number of cases of patent ductus arteriosus was likely inflated because there was no control for low-birth-weight infants. The presentation was followed by a reception for the legislators whom the March of Dimes had lobbied earlier that day.

Following the meeting, I developed a plan to use data from our Hospital Discharge Data System linked with the Birth Certificate Data System to identify birth defect cases. This provided a statewide population-based assessment. We did the extractions and linkages for a one-year birth cohort, the same year's data that were used in the pilot study. At the next March of Dimes annual meeting we presented the overall state data, along with a comparison of the regional pilot study data and the linked data. Once again there was a lot of give and take, and it was readily apparent that the linked data were more complete and accurate. With the birth certificate linkages, we also had considerable data on the characteristics and conditions of the birth population, the denominator for the calculation of strata-specific prevalence estimates. Once again the presentation was followed by a reception with the legislators. A year later a number of the legislators who attended the reception voted to provide funding for our plan to establish a statewide birth defects surveillance program. The data were not perfect, but they clearly had value.

In the meantime, the single-year data were also submitted for the NBDPN annual report and presented at the opening of a state American College of Obstetricians and Gynecologists (ACOG) meeting. The ACOG meeting was formal with a PowerPoint presentation and the audience, primarily physicians and nurses, was very interested and inquisitive. The data showed specific birth defects rates that appeared high relative to national rates and differences among regions of the state. Much of the discussion following the presentation was on the possible reasons for the observed differences. Some of the hypotheses involved potential artifacts in the data, whereas others involved regional differences in behaviors and populations. Once again the interaction was informative for the presenter as well as the audience.

Subsequently we have given presentations at two American Public Health Association annual meetings; one presentation focused on a plan to evaluate the hospital discharge data, using active case/control reviews, and the other on risk factor analyses using the linked birth certificate and hospital discharge data. To date the program has compiled six years of population-based statewide surveillance data using the linked birth-hospital discharge data and two years of active

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case/control reviews. A linkage of the two data sets and their evaluation should be completed soon and will likely provide greater depth and information than any of the previous presentations. The key point is that each of the above-mentioned data sets had both informative and intrinsic value when presented to the appropriate audience, along with clear warnings regarding the data's potential limitations.

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11.6 References

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Appendix 11.1 Data Suppression

Appendix 11.1 Data Suppression

Specific birth defects are often rare events (sometimes extremely rare) leading to a set of issues that must be considered when presenting birth defects data. The public health professional must balance the potentially conflicting goals of information dissemination with protection of the privacy of persons in the community. When the number of cases in a diagnostic category within a group or stratum (such as race or sex) is small or the population from which the cases are determined is small, the risk of allowing a specific individual to be identified may be deemed too large to be acceptable. In such cases, steps must be taken to protect an individual's privacy.

The most common method of preventing the identification of specific individuals in tabular data is through cell suppression. This means not providing counts in individual cells where doing so would potentially allow identification of a specific person. Cell suppression can also be done by combining cells from different small groups to create larger groupings that reduce the risk of identifying individuals. While there are also more sophisticated data perturbation methods that use statistical noise to mask sensitive information, these are generally more suitable for use with economic or financial data than with public health data. This appendix reviews the basic methods, issues, strengths, and vulnerabilities of cell suppression. In addition to protecting privacy, prevalence information is often suppressed when concerns exist regarding possible statistical unreliability of estimates that are based on small numbers.

Suppression Criteria

The first question is whether or not to suppress. Surveillance program administrators and technical staff should be aware that standards used to suppress data may already be set in state laws or in departmental or institutional rules and regulations. It is the responsibility of surveillance staff and administrators to be aware of these standards and practice within their limits. If standards are not established, it behooves a surveillance program to establish rules that will be followed consistently. This is best accomplished with the assistance of an advisory committee, an institutional review or privacy board, or a similar body.

Suppression rules are typically based on a predetermined criterion for the number of diagnosed cases and/or the number of births in the population or subpopulation from which the cases were identified. These numbers may also be thought of as the numerator and the denominator, respectively, of a prevalence estimate. Generally, suppression rules focus on the size of either the numerator or the denominator, the ratio of the numerator to the denominator, or the difference between the numerator and denominator. However the values that trigger suppression vary greatly from one institution or place to another, and there are no set standards. In practice, the rules used vary from relatively liberal to very conservative. Suppression rules for some of the population-based data systems used to assess progress toward the Healthy People 2010 objectives are presented in Table A11.1-1.

Table A11.1-1 Data suppression rules for population-based data systems in the HP2010

Data System	Suppression Criteria				
HIV/AIDS Surveillance System	< 4 cases				
National Notifiable Diseases Surveillance System	Race and Hispanic origin if < 4 cases				
STD Surveillance System	County: < 4 cases; State: < 6 Cases; National: None				

Source: Klein et al., 2002

Each of these suppression criteria is based on simple case counts, but they vary in terms of whether the suppression is of the overall counts or by substrata such as race/ethnicity or geography. In contrast, some surveillance programs will not report data on a birth defect if the case count is less than 5, regardless of the population size, whereas many regularly report single cases. When evaluating the prevalence of birth defects or investigating potential birth defects clusters, it is often necessary to consider birth populations that may consist of small numbers of births. In this situation information on individual cases may be essential to fulfill some of the program's public health functions but should not be included in formal reports.

While reporting small numbers of cases may threaten privacy, the threat may be greatest when reporting from small populations or when the difference between the number of cases and the population count is small. This has led to suppression rules that assess the difference between the prevalence numerator and the denominator or the case count and the population size (e.g., Land, 2001). For example, given the suppression criteria requiring a minimum difference of 15 and a single case of anencephalus in a birth population of 16, the denominator minus the numerator rule would allow the data to be shown. However, in a birth population of 15 the same data would not be shown. Given the nature of anencephalus, an alternative relevant event-specific denominator may be infant deaths in the population. In that case with a birth population of 16, a single anencephalus case would not be shown unless all the infants had died. Thus, even the seemingly simple question of the relevant population to be considered may not be straightforward and should be considered carefully in deciding when to suppress.

Extent of Suppression

Having made the decision to suppress, the question becomes what and how to suppress. The solution that provides the greatest protection of privacy is to suppress an entire table whenever a single cell presents a threat, whereas the solution that provides the least protection is to suppress a single offending cell or only those cells deemed sensitive. Suppressing only sensitive cells is called *primary suppression*. However, when a single cell is suppressed, if column and row totals are provided, they can be used to compute the value of the suppressed cell. Similarly, suppressing multiple cells may allow the values of many or all of the suppressed cells to be revealed through a series of simple arithmetic solutions. This leads some agencies to practice *complementary suppression*, also referred to as *secondary suppression*, in which nonsensitive cells are suppressed in order to support the suppression of sensitive cells. If not properly done, however, the values or approximate ranges of cells in tables created with complementary suppression can also be obtained through the application of simultaneous equations (Geissing, 2001). Complex computer algorithms can be used to determine what cells must be suppressed in order to protect sensitive information. However, these algorithms are not always effective and become excessively complex in large tables (Duncan et al., 2001). One also confronts the issue of increasing data loss when large numbers of cells are used in complementary suppression.

Threat of External Data

A final issue to be considered in deciding when and how to suppress sensitive information is the potential availability of data in multiple tables. It is not enough to simply evaluate the present table with its columns and rows; one must also consider the possible availability of complementary tables. This is especially true in the era of web-based interactive information systems that generate tables for custom queries on demand. Consider a hypothetical case where, in the process of creating a table for an annual report, it was determined that cells showing pyloric stenosis counts for the black population were potentially sensitive and the decision was made to provide only the total number of cases. Subsequently it is determined that effectively suppressing the black population's case counts would require

Appendix 11.1 A11.1-2 Data Presentation

complementary suppression of the white population's case counts. Given that the white population's data were not sensitive, they may be subsequently published in a separate table. If so, the resulting data could be combined with the original table in order to reveal the black population's data. A similar situation would arise if, to protect privacy and present all of the data, the population strata were collapsed and subsequently data for one of the strata were published.

Summary on Suppression

The more restrictive a suppression rule, the less information a given table or report will provide. The weaker a suppression rule, the greater the potential threat of revealing confidential health information. It is a question of balancing the threat to individual privacy with the public health value of presenting the data. Overall, deciding when and how to suppress birth defects information is more a social, political, and legal issue than a technical one. The technical aspects are quite straightforward, but the contextual and procedural/policy issues are likely not to be. These all need to be considered and balanced in the local context before informed decisions can be made to suppress or not to suppress data in program reports or other documents.

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See also American Statistical Association: http://www.amstat.org/comm/cmtepc/index.cfm?fuseaction=1. Accessed 5/21/08.

Appendix 11.2

Use of Geographic Information Systems (GIS) to Map Data

Appendix 11.2 Use of Geographic Information Systems (GIS) to Map Data

The application of Geographic Information Systems (GIS) methods has become an integral component of aggregating, analyzing, and evaluating health data. The current practical applications of GIS in epidemiologic studies range from descriptive statistics (i.e., plotting data on a map) to evaluation of spatial relations between environmental exposures and health outcomes.

Several definitions exist for geographic information systems. One of the most recent, as found in *Healthy People 2010*, defines GIS as "powerful tools combining geography, data and computer mapping" (U.S. Department of Health and Human Services, 2000). Software packages available today, such as ArcMap and MapInfo, integrate many GIS functions. These include (1) database management, (2) data manipulation and analysis, and (3) data presentation (i.e., displaying data on a map). To be included in GIS, the data should have some kind of geographical or spatial component that can be translated into digit maps.

Digital Map Formats

GIS applications use either a vector or a raster map format, or a combination of the two. In *vector* maps (Figure A11.2-1) geographic features are represented by points (e.g., location of infants with birth defects), lines (e.g., streets), and polygons (e.g., census tracts) (Rogers, 1999). These features are based on latitude and longitude coordinates of the different objects. The vector format is the most commonly used in public health. In *raster* maps the data are stored as digital images (e.g., orthophotos, scanned maps) (Vine et al., 1997). Usually a grid cell is used to represent a feature, and these cells can be connected. As such, smaller cells provide a more detailed resolution. Obtaining quality maps for a given geographical area for the time period of interest is crucial as maps are static while environments change.

Bringing Health Data into GIS

Ultimately, the application of GIS to birth defects data requires the transformation, as accurately as possible, of health records containing addresses or location information into geographic objects. This process is called *geocoding*, also known as address matching. During geocoding, latitude and longitude coordinates are assigned by the GIS software to each address by matching against an *address-range* (i.e., street segment) in a street reference map such as the Census Topologically Integrated Geographic Encoding and Referencing (TIGER) files (Croner et al., 1996). Interpolation is used to estimate the actual address location within the given range (Rushton, 1999). The address match rate depends on several factors, including the completeness of addresses in health records and the accuracy of reference maps (McElroy et al., 2003). In case of incorrect or missing house numbers and/or street names, coordinates are usually assigned to a centroid of a larger geographical entity, such as a census tract or a ZIP code. If available, other reference files such as tax parcel databases can also be used for geocoding purposes. Alternatively, in areas where latitude and longitude coordinates have not been predetermined (e.g., rural communities), a global positioning system (GPS) device can be used, although this may prove time and resource demanding. Figure A11.2-2 shows an example of how a point is placed within an address range.

Figure A11.2-1 An Example of GIS Data Layers

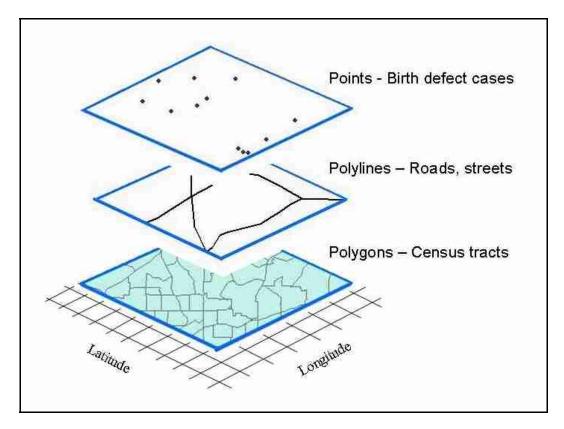
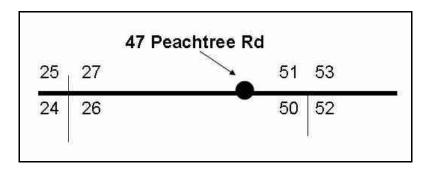


Figure A11.2-2 A Specific Location Within an Address Range



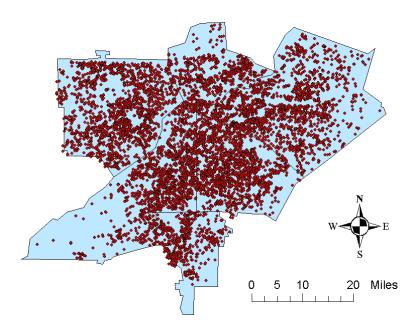
Mapping Data

Once health data are brought into a GIS database, users need to be aware of several important issues in data mapping. For example, different spatial databases must have the same scale and projection (McLafferty and Cromley, 1999). Otherwise data will be distorted or cannot be mapped together. *Map scale* shows the relationship between a unit of length on a map and the corresponding length on the ground. It is also an expression of how much the area represented has been reduced on the map. The smaller the scale, the larger the area displayed on a map. *Map projections* are attempts to portray/transform the surface of the three-dimensional earth or a portion of the earth on a flat map using a mathematical model. Some distortions of conformality, distance, direction, scale, and area always result from this process. Maps that focus on maintaining one feature (e.g., preserving distance) must distort other features (e.g., area, shape). Maps that accurately reflect area are called *equal-area maps*, while maps that correctly show the distance between points are called *equidistant maps*.

Two types of maps frequently used in public health research are dot-density and choropleth maps (Rogers, 1999).

Dot-density mapping. *Dot-density maps* are the simplest way to display events. These maps use dots or other symbols to represent the number of occurrences of a given data characteristic (Thrall, 1999). Each dot or symbol used on the map may represent a single entity (one dot = one case) or a group (one dot = 1,000 people). Dot-density maps are useful for area comparisons. However, dot-density maps need to be interpreted with caution regarding the "symbol to data characteristic" ratio. It is also important to keep in mind that dots do not always indicate the exact location of the data. An example of a dot-density map for metropolitan Atlanta is presented in Figure A11.2-3 (Source: Siffel et al., 2006, Figure 1, p. 828).



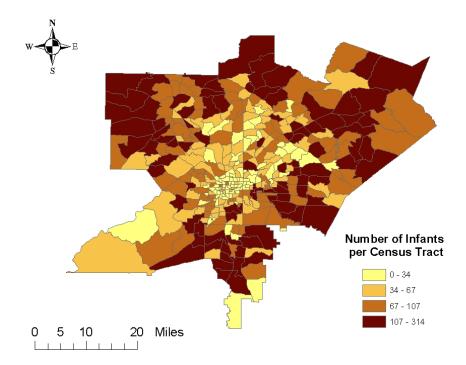


Appendix 11.2 A11.2-3 Data Presentation

Choropleth mapping. Choropleth maps are area maps in which polygons (e.g., census tracts, counties) are shaded, colored, or patterned according to the extent to which a given attribute (such as population size or disease rate) is associated with each polygon. Choropleth maps are also called *thematic maps* or *shaded maps*. An example of a choropleth map for metropolitan Atlanta is presented in Figure A11.2-4 (Source: Siffel et al., 2006, Figure 2, p. 828).

It is important to choose the right characteristics for map presentations as the choice of color, pattern, size, polygon shape, and class intervals can impact how one interprets the information presented in a map. Single-color maps with varying color intensity (shades) are often an effective means of presenting data, but the use of differing patterns can help a black-and-white or grey-scale map. Similar-size polygons are recommended to the extent possible, as a few large polygons can dominate a map, leading to misinterpretation of information. Proportions or rates can be displayed by different class interval schemes, such as equal intervals (equal ranges of values) or quintiles (equal number of polygons falling into each class defined by dividing the range of values). The latter method is particularly useful for presenting skewed data. These methods are standard in GIS software.

Figure A11.2-4 Choropleth Map of Infants per Census Tract in Metropolitan Atlanta, 1990



Additional Technical Details

Maps showing point locations or even aggregate data in a small geographic area have the potential to reveal the identity of individuals (Cox, 1996). Therefore, as noted elsewhere in this chapter (see Appendix 11.1 on Data Suppression), one must generally limit the presentation of disaggregated birth defects information. While GIS methods and techniques exist for protecting privacy and limiting disclosure of information by geographically masking individual records (Armstrong et al., 1999), the use of masked data in small-area analysis can limit one's ability to detect clusters of cases (Kamel Boulos et al., 2005). As such, careful choice of geographical units and data aggregation are vital.

Below we present several practical suggestions for preparing and presenting maps above and beyond those already mentioned.

- The use of the same scale, colors, class intervals, and legends when presenting a series of maps.
- The inclusion of a scale bar and a "North" arrow.
- The use of patterns when printing in black and white. Color maps produced on a black-and-white printer usually do not provide as good results as grey scale.
- Avoid the use of red and green on the same map.
- ➤ Be wary of font-related problems. If symbols, which are special GIS fonts, are used on a map, do not export the map as an MS Windows meta file (.wmf). This type of file requires access to the GIS fonts. Similarly, do not include such files in presentations being made on an unfamiliar computer. If the GIS fonts are not available, other fonts will be substituted for symbols in the image. Instead, export your maps as JPEG files.

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Appendix 11.3 Data Users Matrix

Appendix 11.3 Data Users Matrix

Sample Questions Asked

Information Needs/Data Presentation Suggestions

Surveillance Program Staff

This group is likely to require process indicators useful for management. Members of this group may notice possible clusters. This audience also needs additional information about denominator issues and data quality.

- How many abstracts were completed per field staff person?
- We have noticed more cases of birth defect x in this hospital; is that unusual?
- Data on labor hours and abstracting rates.
- Data on birth prevalence, usually in comparison to some standard, such as the entire state.
- Internal exhibits in terms of surveillance parameters: Completeness/Ascertainment, Case Processing Times.

Researcher

This audience is likely to be interested in:

- Descriptive epidemiology (describing occurrence of birth defects by person, place, and time).
- Analytic epidemiology (finding causes of birth defects).
- Obtaining birth defect cases for related studies.
- Methodological issues.
- Availability of data on individual types of birth defects and on cases, both identified and deidentified, for ecologic studies and etiologic research.
- What are the patterns of birth defect occurrence by person, place, and time?
- What is associated with risk of birth defect x?
- How many cases of specific birth defects are represented in the database of the surveillance program?
- How do I get access to records for persons included in the surveillance program for research studies?
- Printed and web reports and charts with routinely tabulated results allow for quick response to many inquiries.
- Specially tabulated results
- Line item data with and without confidential information.
- Interactive web-based tool for custom queries allows for easy access and reduces staff time in responding to simple routine and non-routine aggregate data requests. See, for example, http://soupfin.tdh.state.tx.us/bdefdoc.htm

Sample Questions Asked

Information Needs/Data Presentation Suggestions

Physician and Other Health Care Providers

This audience is often interested in outcomes (pregnancy, how does the child do, survival/treatment), as well as birth defect prevalence. Members of this group may notice possible clusters. This audience also needs additional information about denominator issues and data quality. Statistical output for this audience may well run more detailed in terms of clinical detail. Allow for the presentation of small cases series. Members of this group often have the skill to interpret tabular data, but do not assume they will necessarily have the statistical sophistication that may be lacking in other audiences.

- Which defects are most common?
- What are the trends in birth defects over time?
- Are there unexpectedly high rates in my area or facility?
- Printed reports.
- Web-based data.

Social/Education Service Provider

Needs from this audience are usually geographic in nature. Birth defects programs can promote use of data among this group by providing maps and other data that respond to these information needs.

- Describe the population we serve in terms of location, income, other variables.
- How many babies with complex congenital heart defects do you estimate will be born during the next five years in the area for which our children's hospital provides clinical care?
- Mapping location of clinics compared to appropriate birth defect cases.
- Time series analyses with projections (e.g., how many children with x syndrome will be born in x county for the next five years in order to project needs for special education teachers, etc.?).

Local Health Department

This audience tends to be interested in epidemiological data. Members of this group may also want to link your data with data they have on environmental concerns (e.g., factories, toxic waste sites). There is a risk of misuse of data if users do not understand the unique aspects of birth defects data.

- What are our rates?
- How do we compare with the rest of the state?
- How do we compare with the nation?
- Are there links between birth defect clusters and local environmental concerns?
- What clusters (in our area) are you dealing with?
- What are the trends in birth defects over time?

- Epidemiologic data provided by zip code, city, county, region.
- Case characteristic summaries as tables, as well as rate tables and graphics, including trends.
 Consider also the presentation of rate ratios.
- Reports of cluster investigations; maps of clusters investigated.
- Have a document written in language accessible to the lay person explaining some of the finer points of interpreting birth defects data.

Sample Questions Asked

Information Needs/Data Presentation Suggestions

Maternal and Child Health Programs

Members of this group might be professionals in Women Infants, and Children (WIC) or Title V programs. They tend to be interested in trends over time, rates, outcomes, and surveillance operations.

- Do you have any information on folic acid education programs that can be targeted to our Hispanic clients?
- Items here will likely address racial disparities and the demographics that underlie differences in race, including cultural health practices and SES.
- GIS/spatial analysis may be relevant here.

Family of Child with Birth Defect(s)

This audience tends to like information about known causes and risk factors for particular birth defects. Members of this group are interested in seeing birth defects data linked with potential teratogens or environmental concerns and the outcomes. They may desire information on educational, social, and clinical services available for children with specific types of birth defects.

- I have a child with a cleft lip. What information do you have on the causes of this condition?
- I just had a baby with spina bifida. What information can you give me about this condition and where can I get specialized care for her?
- Do you have any information on support groups in my community for parents of children with Down syndrome?
- What caused my child's birth defect?
- Have there been clusters investigated in my area?

- Literature summaries.
- Maps of cluster investigations.
- Charts comparing local/state rates to other areas.
- Be sure to explain the difference between individual- and population-level information

Students (public health, medical, nursing, allied health, or other college/university)

Needs among this audience might include:

- o General education (e.g., lectures to a class)
- Specific education (e.g., practicum placements)
- Research (e.g., data and guidance on papers, theses, dissertations)

You might present to these types of students during Grand Rounds or at local seminars or conferences. They may also submit specific requests as a result of papers or projects they are working on.

- Can you provide me with information about changes in the occurrence of neural tube defects in ____ following the fortification of cereal grains with folic acid?
- Printed reports.
- Copies of manuscripts.
- Interactive web-based tool for custom queries allows for easy access and reduces staff time in responding to simple routine and non-routine aggregate data requests.
- Raw data.

Sample Questions Asked

Information Needs/Data Presentation Suggestions

News/Media Person

Media personnel are generally interested in comparative rates region/state/nation. Also, they will likely need information about causes and risk factors. Their questions may or may not relate to a specific community concern or cluster. They tend to request large amounts of data and use very little of it.

- What is the cause of x birth defect?
- Are the cases of x birth defect linked with the toxic dump, military base, factory, vaccine, etc?
- Printed and web reports and charts with routinely tabulated results allow for quick response to many inquiries.
- Have a document written in language accessible to the lay person explaining some of the finer points of working with and interpreting birth defects data.

Legislator/Policy Maker

This type of information request usually comes from higher up in the agency or from advocacy groups. Top information needs include cost of program, cost of birth defects to the state, number of people served by the program, and staffing data. This audience would be interested in surveillance data connected to other information such as regional variation or costs. Members of this group may also request administrative data—improvements in program efficiency, budget information, increases in caseload (live births, hospitals).

- How many staff (FTEs) does the surveillance program employ?
- What is the cost of the program?
- How many babies with birth defects are born each year in my legislative district?
- What are the estimated lifetime costs of caring for a child with spina bifida?
- What is the impact of this policy? (e.g., decrease in rates of NTDs after fortification of food supply with folic acid)

- Narrative reports.
- Graphs of birth prevalence over time (for before/after comparisons) or comparing communities with different policies.
- Statistical exhibits intended for this audience should address not only descriptive aspects, but also the quantitative burden of disease.

Advocacy Group

The needs of this group will vary depending on what they are advocating for. This audience will need exhibits at two levels—exhibits for the lay public and exhibits for policy makers—and the distinction needs to be clear.

- What are the rates in z location for birth defect x?
- Why are you not collecting data on birth defect x?
- What is the cost to y unit of government for treating birth defect x?
- Cost, magnitude, impact.
- Birth defects data linked with environmental data.

Sample Questions Asked Other Community Members Members of this group usually contact the registry to report concern of a possible cluster. They are frequently interested in environmental exposures and birth defects. What are the rates of birth defects in my community? Are birth defects higher here than elsewhere? If higher, can the excess birth defects be linked to environmental concerns?

Appendix 11.4 What Type of Chart or Graph Should I Use?

Appendix 11.4 What Type of Chart or Graph Should I Use?

Research illuminating human perceptions of graphical representations offers us clues as to how to select the best representation for a given type of data. Below we present examples of the most common types of graphs and charts, along with suggestions on when they might be used.

Pie charts can be effective for communicating simple proportions (see Figure A11.4-1). When comparing several proportions, convention dictates that none of the radii should be at the "12 o'clock" position (Hollands, 2003). Pie charts do not need legends, instead the series name and percentage should be positioned next to the appropriate slice.

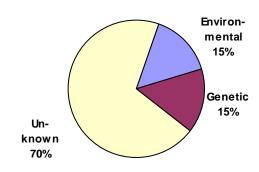


Figure A11.4-1 What causes birth defects?

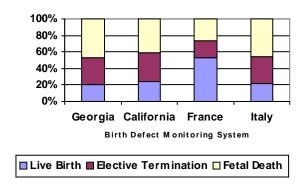
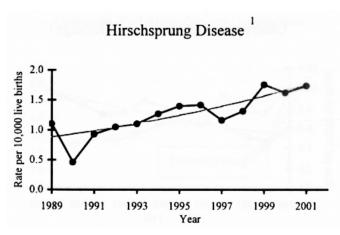


Figure A11.4-2 Pregnancy Outcomes, Down Syndrome

A 100% stacked bar chart can be used to compare proportions between two or more data sets (see Figure A11.4-2). However, be careful about presenting too many data points as the graph may become too busy to convey information effectively. An alternative would be a series of several pie charts, although 100% stacked bars allow for more consistent comparisons.



Time series are nearly always demonstrated using a *line chart*, with a marker at each year. Figure A11.4-3 includes a regression line indicating that the change in rates is indeed statistically significant, thereby adding important information to this chart.

¹Trend is significant; details are given in Table 3

Figure A11.4-3 Rates of Hirschsprung disease 1989–2001

Certainly the most common need for graphical representations of birth defects data is the comparison of rates of cases among persons and places. *Bar charts* are an ideal choice for this because they give an impression of relative differences but, unlike line charts, do not give the impression that moving left-to-right is a time progression (see Figure A11.4-4). (Note: This chart also demonstrates the use of white breaks in the bars in lieu of gridlines across the whole plot area.)

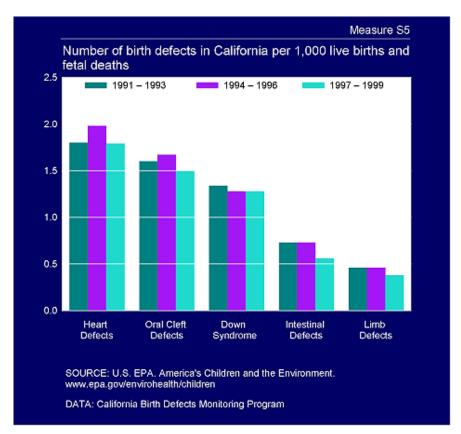


Figure A11.4-4 Bar chart demonstrating cases per 1,000 live births

Bar charts can also be used to convey information about the statistical significance of rates by using drop lines to represent confidence limits, as in Figure A11.4-5. Note: the best way to ensure that confidence limits are represented correctly is to import the results directly into your graphic software from your analysis software. However, it is also possible to produce the irregular confidence limits found when using Poisson regression in Microsoft Office products (see the document "Plotting Irregular 95% Confidence Intervals" on the Members Only section of the NBDPN website).

Turner Syndrome 1999-2004

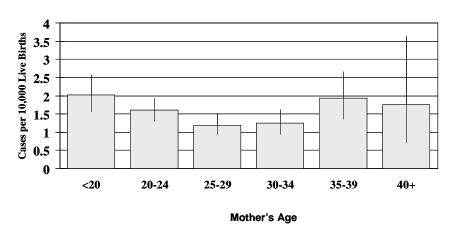
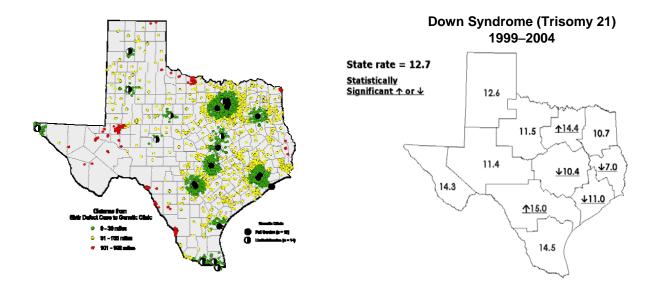


Figure A11.4-5 Bar chart with confidence limits

Figure A11.4-6 Spot Map

Figures A11.4-6 and A11.4-7 below, respectively, present examples of maps of epidemiological data. For further detail on the use of Geographic Information Systems see Appendix 11.2.



Appendix 11.4 A11.4-3 Data Presentation

Figure A11.4-7 Area Map

When places are not contiguous or for some other reason would be difficult to display on a map, a bar chart such as Figure A11.4-8 would be suitable.

Omphalocele by State, 1999-2003

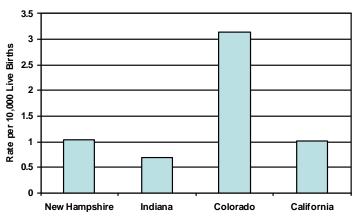


Figure A11.4-8 Displaying geographic data with a bar chart

When only the general place rather than a specific site is relevant (e.g., entire state versus specific regions or locales within the state), it is possible to use an area map (see Figure A11.4-9).

BRFSS Maps

Year - 2006

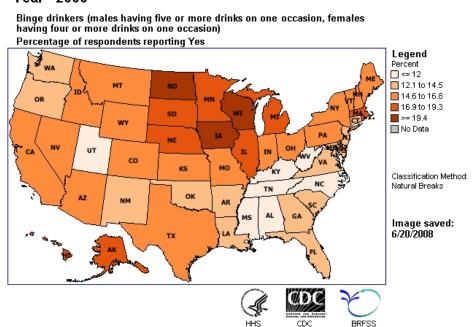


Figure A11.4-9 Displaying geographic data in an area map

In Table A11.4-1 below you will find one scheme for selecting the appropriate graphic representation given the type of data you will be presenting.

Before making your final decision, however, you should also ask yourself two questions that relate less to the nature of your data and more to your own personal preferences and the needs/interests of your audience:

- Am I comfortable explaining this graph? If the answer is no, find an alternative format with which you are more comfortable.
- ➤ Given my audience, should I sacrifice detail for clarity, or clarity for detail? For example, an audience of foster parents would probably benefit from clarity, whereas an audience of epidemiologists will readily comprehend your meaning and will rather be looking for additional detail about methods or sample characteristics.

Table A11.4-1 Selecting a method of illustrating epidemiologic data (adapted from *Principles of Epidemiology*, 3rd edition, U.S. Department of Health and Human Services).

If Data Are:		And These Conditions Apply:		Then Choose:
Proportions		< 6 data points	1 series	Pie chart (Sample Figure 4)
			>1 series	100% stacked bars
				(Sample Figure 5)
		6+ data points	1+ series	Consider combining data point
				categories or table.
				(Sample Figure 5)
Time Series		Numbers of Cases		Line chart (Sample Figure 6)
Data with				Bar chart (Sample Figures 7, 8)
discrete categories				
Place	Number of	Not readily identified on map		Bar chart (Sample Figure 11)
	cases	Readily identified	Specific site	Spot map (Sample Figure 9)
		on map	important	
			Specific site	Area map (Sample Figure 10)
			unimportant	
	Rates			Area map (Sample Figure 12)

Cited References on Graphic Presentation

Hollands JG. The classification of graphical elements. *Canadian Journal of Experimental Psychology*. 2003;57(1):38-47.

Centers for Disease Control and Prevention, Office of Workforce and Career Development. *Principles of Epidemiology: An Introduction to Applied Epidemiology and Biostatistics.* 3rd edition. Atlanta, GA: U.S. Department of Health and Human Services, Centers for Disease Control and Prevention; n.d.

Appendix 11.4 A11.4-5 Data Presentation